

Supplementary materials

Detecting *MUC1* variants among clinicopathologically diagnosed ADTKD patients

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Supplementary Table S1. The gene list constructed using HaloPlex (version 2, 128 genes)

<i>ACE</i>	<i>BMP7</i>	<i>CXCL12</i>	<i>FZD4</i>	<i>IQCB1</i>	<i>NPHS2</i>	<i>RET</i>	<i>TBX18</i>	<i>WNT4</i>
<i>ACTN4</i>	<i>CC2D2A</i>	<i>CXCR4</i>	<i>FZD8</i>	<i>ITGA8</i>	<i>NUP107</i>	<i>ROBO2</i>	<i>TMEM138</i>	<i>WNT5A</i>
<i>ADCK4</i>	<i>CD2AP</i>	<i>DCDC2</i>	<i>GATA3</i>	<i>KAL1</i>	<i>NUP133</i>	<i>ROR1</i>	<i>TMEM231</i>	<i>WNT7A</i>
<i>AGT</i>	<i>CDC5L</i>	<i>DSTYK</i>	<i>GDNF</i>	<i>KIAA0586</i>	<i>NXF5</i>	<i>ROR2</i>	<i>TMEM237</i>	<i>WNT7B</i>
<i>AGTR1</i>	<i>CEP164</i>	<i>EP300</i>	<i>GLIS2</i>	<i>LAMB2</i>	<i>OFD1</i>	<i>RPGLIP1L</i>	<i>TMEM67</i>	<i>WNT9B</i>
<i>AGTR2</i>	<i>CEP290</i>	<i>EYA1</i>	<i>GREM1</i>	<i>LMNA</i>	<i>OSR1</i>	<i>SALL1</i>	<i>TNXB</i>	<i>WT1</i>
<i>AHI1</i>	<i>CEP41</i>	<i>FAT3</i>	<i>GRIP1</i>	<i>LMX1B</i>	<i>PAX2</i>	<i>SDCCAG8</i>	<i>TP53</i>	<i>ZEB2</i>
<i>ALG13</i>	<i>CEP83</i>	<i>FAT4</i>	<i>HNF1B</i>	<i>MDM2</i>	<i>PAX8</i>	<i>SIX1</i>	<i>TRAP1</i>	<i>ZNF423</i>
<i>ANKS3</i>	<i>CHD1L</i>	<i>FGF2</i>	<i>HOXA13</i>	<i>MKKS</i>	<i>PDE6D</i>	<i>SIX2</i>	<i>TRPC6</i>	
<i>ANKS6</i>	<i>CHD4</i>	<i>FGF20</i>	<i>HOXD11</i>	<i>MUC1</i>	<i>PKD1</i>	<i>SIX5</i>	<i>TTC21B</i>	
<i>ANLN</i>	<i>CHRM3</i>	<i>FGF9</i>	<i>IFN2</i>	<i>MYO1E</i>	<i>PKD2</i>	<i>SLIT2</i>	<i>UMOD</i>	
<i>APOL1</i>	<i>CITED1</i>	<i>FOXD1</i>	<i>IFT172</i>	<i>NEK8</i>	<i>PKHD1</i>	<i>SOX17</i>	<i>UPK3A</i>	
<i>ARL13B</i>	<i>COQ6</i>	<i>FRAS1</i>	<i>IFT81</i>	<i>NPHP1</i>	<i>PLCE1</i>	<i>SPRY1</i>	<i>VANGL2</i>	
<i>BMP2</i>	<i>CSPP1</i>	<i>FREM1</i>	<i>INPP5E</i>	<i>NPHP3</i>	<i>PODXL</i>	<i>SRGAP1</i>	<i>WDR19</i>	
<i>BMP4</i>	<i>CTDNEP1</i>	<i>FREM2</i>	<i>INVS</i>	<i>NPHP4</i>	<i>REN</i>	<i>TBX1</i>	<i>WNT11</i>	

Supplementary Table S2. The gene list constructed using HaloPlex (version 4, 172genes)

<i>ACE</i>	<i>B9D1</i>	<i>CD2AP</i>	<i>CTDNEP1</i>	<i>FOXD1</i>	<i>IFN2</i>	<i>LMNA</i>	<i>NUP107</i>	<i>RET</i>	<i>TBX1</i>	<i>TSC1</i>	<i>WNT7B</i>
<i>ACTN4</i>	<i>B9D2</i>	<i>CDC5L</i>	<i>CXCL12</i>	<i>FRAS1</i>	<i>IFT27</i>	<i>LMX1B</i>	<i>NUP133</i>	<i>ROBO2</i>	<i>TBX18</i>	<i>TSC2</i>	<i>WNT9B</i>
<i>ADCK4</i>	<i>BBIP1</i>	<i>CENPF</i>	<i>CXCR4</i>	<i>FREM1</i>	<i>IFT43</i>	<i>LZTFL1</i>	<i>NXF5</i>	<i>ROR1</i>	<i>TCTN2</i>	<i>TTC8</i>	<i>WNT11</i>
<i>AGT</i>	<i>BBS1</i>	<i>CEP41</i>	<i>DCDC2</i>	<i>FREM2</i>	<i>IFT81</i>	<i>MDM2</i>	<i>OFD1</i>	<i>ROR2</i>	<i>TCTN3</i>	<i>TTC21B</i>	<i>WT1</i>
<i>AGTR1</i>	<i>BBS2</i>	<i>CEP83</i>	<i>DDX59</i>	<i>FZD4</i>	<i>IFT122</i>	<i>MKKS</i>	<i>OSR1</i>	<i>RPGLIP1L</i>	<i>TMEM67</i>	<i>UMOD</i>	<i>XPNPEP3</i>
<i>AGTR2</i>	<i>BBS4</i>	<i>CEP104</i>	<i>DSTYK</i>	<i>FZD8</i>	<i>IFT140</i>	<i>MKS1</i>	<i>PAX2</i>	<i>SALL1</i>	<i>TMEM138</i>	<i>UPK3A</i>	<i>ZEB2</i>
<i>AHI1</i>	<i>BBS5</i>	<i>CEP120</i>	<i>DYNC2H1</i>	<i>GANAB</i>	<i>IFT172</i>	<i>MUC1</i>	<i>PAX8</i>	<i>SDCCAG8</i>	<i>TMEM216</i>	<i>VANGL2</i>	<i>ZNF423</i>
<i>ALG13</i>	<i>BBS7</i>	<i>CEP164</i>	<i>EP300</i>	<i>GATA3</i>	<i>INPP5E</i>	<i>MYO1E</i>	<i>PDE6D</i>	<i>SIX1</i>	<i>TMEM231</i>	<i>WDPCP</i>	
<i>ALMS1</i>	<i>BBS10</i>	<i>CEP290</i>	<i>EYA1</i>	<i>GDNF</i>	<i>INVS</i>	<i>NEK1</i>	<i>PKD1</i>	<i>SIX2</i>	<i>TMEM237</i>	<i>WDR19</i>	
<i>ANKS3</i>	<i>BBS12</i>	<i>CHD1L</i>	<i>FAT3</i>	<i>GLIS2</i>	<i>IQCB1</i>	<i>NEK8</i>	<i>PKD2</i>	<i>SIX5</i>	<i>TNXB</i>	<i>WDR34</i>	
<i>ANKS6</i>	<i>BMP2</i>	<i>CHD4</i>	<i>FAT4</i>	<i>GREM1</i>	<i>ITGA8</i>	<i>NODAL</i>	<i>PKHD1</i>	<i>SLIT2</i>	<i>TP53</i>	<i>WDR35</i>	
<i>ANLN</i>	<i>BMP4</i>	<i>CHRM3</i>	<i>FGF2</i>	<i>GRIP1</i>	<i>KAL1</i>	<i>NPHP1</i>	<i>PLCE1</i>	<i>SOX17</i>	<i>TRAF3IP1</i>	<i>WDR60</i>	
<i>APOL1</i>	<i>BMP7</i>	<i>CITED1</i>	<i>FGF9</i>	<i>HOXA13</i>	<i>KIAA0586</i>	<i>NPHP3</i>	<i>PODXL</i>	<i>SPRY1</i>	<i>TRAP1</i>	<i>WNT4</i>	
<i>ARL6</i>	<i>C5orf42</i>	<i>COQ6</i>	<i>FGF20</i>	<i>HOXD11</i>	<i>KIF14</i>	<i>NPHP4</i>	<i>PTHB1</i>	<i>SRGAP1</i>	<i>TRIM32</i>	<i>WNT5A</i>	
<i>ARL13B</i>	<i>CC2D2A</i>	<i>CSPP1</i>	<i>FGFR2</i>	<i>HNF1B</i>	<i>LAMB2</i>	<i>NPHS2</i>	<i>REN</i>	<i>TBC1D1</i>	<i>TRPC6</i>	<i>WNT7A</i>	

Supplementary Table S3. The gene list constructed using HaloPlex (version 5, 159 genes)

<i>ACE</i>	<i>BBS1</i>	<i>CEP41</i>	<i>DYNC2H1</i>	<i>GANAB</i>	<i>IFT80</i>	<i>LMNA</i>	<i>PDE6D</i>	<i>SIX1</i>	<i>TMEM107</i>	<i>WDPCP</i>
<i>AGT</i>	<i>BBS2</i>	<i>CEP83</i>	<i>DZIP1L</i>	<i>GATA3</i>	<i>IFT81</i>	<i>LMX1B</i>	<i>PIBF1</i>	<i>SIX2</i>	<i>TMEM138</i>	<i>WDR19</i>
<i>AGTR1</i>	<i>BBS4</i>	<i>CEP104</i>	<i>EP300</i>	<i>GDNF</i>	<i>IFT122</i>	<i>LZTFL1</i>	<i>PKD1</i>	<i>SIX5</i>	<i>TMEM216</i>	<i>WDR34</i>
<i>AGTR2</i>	<i>BBS5</i>	<i>CEP120</i>	<i>EVC</i>	<i>GLIS2</i>	<i>IFT140</i>	<i>MKKS</i>	<i>PKD2</i>	<i>SLIT2</i>	<i>TMEM231</i>	<i>WDR35</i>
<i>AHI1</i>	<i>BBS7</i>	<i>CEP164</i>	<i>EVC2</i>	<i>GLIS3</i>	<i>IFT172</i>	<i>MKS1</i>	<i>PKHD1</i>	<i>SOX17</i>	<i>TMEM237</i>	<i>WDR60</i>
<i>ALG9</i>	<i>BBS10</i>	<i>CEP290</i>	<i>EXOC4</i>	<i>GRIP1</i>	<i>INPP5E</i>	<i>MUC1</i>	<i>PTHB1</i>	<i>SPRY1</i>	<i>TNXB</i>	<i>WNT4</i>
<i>ALMS1</i>	<i>BBS12</i>	<i>CHD1L</i>	<i>EXOC8</i>	<i>GRLF1</i>	<i>INVS</i>	<i>NEK1</i>	<i>REN</i>	<i>SRGAP1</i>	<i>TRAF3IP1</i>	<i>WT1</i>
<i>ANKS3</i>	<i>C2CD3</i>	<i>CHD4</i>	<i>EYA1</i>	<i>HNF1B</i>	<i>IQCB1</i>	<i>NEK8</i>	<i>RET</i>	<i>TBC1D1</i>	<i>TRIM32</i>	<i>XPNPEP3</i>
<i>ANKS6</i>	<i>C5orf42</i>	<i>CHRM3</i>	<i>FAN1</i>	<i>HOXA13</i>	<i>ITGA8</i>	<i>NPHP1</i>	<i>ROBO2</i>	<i>TBC1D32</i>	<i>TSC1</i>	<i>ZNF423</i>
<i>ARL6</i>	<i>C21orf2</i>	<i>CITED1</i>	<i>FGF9</i>	<i>HPRT1</i>	<i>JAG1</i>	<i>NPHP3</i>	<i>RPGLIP1L</i>	<i>TBX1</i>	<i>TSC2</i>	
<i>ARL13B</i>	<i>CC2D2A</i>	<i>CSPP1</i>	<i>FGF20</i>	<i>HYLS1</i>	<i>KAL1</i>	<i>NPHP4</i>	<i>SALL1</i>	<i>TBX18</i>	<i>TTBK2</i>	
<i>ATXN10</i>	<i>CCDC28B</i>	<i>CTDNEP1</i>	<i>FGFR2</i>	<i>ICK</i>	<i>KIAA0586</i>	<i>OFD1</i>	<i>SARS2</i>	<i>TCTN1</i>	<i>TTC8</i>	
<i>B9D1</i>	<i>CDC5L</i>	<i>DCDC2</i>	<i>FRAS1</i>	<i>IFN2</i>	<i>KIF7</i>	<i>PAX2</i>	<i>SCLT1</i>	<i>TCTN2</i>	<i>TTC21B</i>	
<i>B9D2</i>	<i>CENPF</i>	<i>DDX59</i>	<i>FREM1</i>	<i>IFT27</i>	<i>KIF14</i>	<i>PAX8</i>	<i>SDCCAG8</i>	<i>TCTN3</i>	<i>UMOD</i>	
<i>BBIP1</i>	<i>CEP19</i>	<i>DSTYK</i>	<i>FREM2</i>	<i>IFT43</i>	<i>LIFR</i>	<i>PBX1</i>	<i>SEC61A1</i>	<i>TMEM67</i>	<i>VANGL2</i>	

Supplementary Table S4. The gene list constructed using HaloPlex (version 6, 164genes)

<i>ACE</i>	<i>BBIP1</i>	<i>CEP83</i>	<i>EP300</i>	<i>GLIS3</i>	<i>IFT140</i>	<i>LZTFL1</i>	<i>PKD1</i>	<i>SOX17</i>	<i>TMEM231</i>	<i>WNT4</i>
<i>ACTG2</i>	<i>BBS1</i>	<i>CEP104</i>	<i>EVC</i>	<i>GREB1L</i>	<i>IFT172</i>	<i>MAPKBP1</i>	<i>PKD2</i>	<i>SPRY1</i>	<i>TMEM237</i>	<i>WT1</i>
<i>AGT</i>	<i>BBS2</i>	<i>CEP120</i>	<i>EVC2</i>	<i>GRIP1</i>	<i>INPP5E</i>	<i>MKKS</i>	<i>PKHD1</i>	<i>SRGAP1</i>	<i>TNXB</i>	<i>XPNPEP3</i>
<i>AGTR1</i>	<i>BBS4</i>	<i>CEP164</i>	<i>EXOC4</i>	<i>HNF1B</i>	<i>INTU</i>	<i>MKS1</i>	<i>REN</i>	<i>SUFU</i>	<i>TRAF3IP1</i>	<i>ZNF423</i>
<i>AGTR2</i>	<i>BBS5</i>	<i>CEP290</i>	<i>EXOC8</i>	<i>HOXA13</i>	<i>INVS</i>	<i>MUC1</i>	<i>RET</i>	<i>TBC1D32</i>	<i>TRIM32</i>	
<i>AHI1</i>	<i>BBS7</i>	<i>CHD1L</i>	<i>EYA1</i>	<i>HPRT1</i>	<i>IQCB1</i>	<i>NEK1</i>	<i>ROBO2</i>	<i>TBX1</i>	<i>TSC1</i>	
<i>ALG8</i>	<i>BBS9</i>	<i>CHD4</i>	<i>FAN1</i>	<i>HYLS1</i>	<i>ITGA8</i>	<i>NEK8</i>	<i>RPGRIP1L</i>	<i>TBX18</i>	<i>TSC2</i>	
<i>ALG9</i>	<i>BBS10</i>	<i>CHRM3</i>	<i>FGF20</i>	<i>INF2</i>	<i>JAG1</i>	<i>NPHP1</i>	<i>SALL1</i>	<i>TCTEX1D2</i>	<i>TTC8</i>	
<i>ALMS1</i>	<i>BBS12</i>	<i>CITED1</i>	<i>FGFR2</i>	<i>IFT27</i>	<i>KAL1</i>	<i>NPHP3</i>	<i>SARS2</i>	<i>TCTN1</i>	<i>TTC21B</i>	
<i>ANKS6</i>	<i>C2CD3</i>	<i>CRB2</i>	<i>FRAS1</i>	<i>IFT43</i>	<i>KIAA0556</i>	<i>NPHP4</i>	<i>SCLT1</i>	<i>TCTN2</i>	<i>UMOD</i>	
<i>ARL6</i>	<i>C5orf42</i>	<i>CSPP1</i>	<i>FREM1</i>	<i>IFT52</i>	<i>KIAA0586</i>	<i>OFD1</i>	<i>SDCCAG8</i>	<i>TCTN3</i>	<i>VANGL2</i>	
<i>ARL13B</i>	<i>C8orf37</i>	<i>DCDC2</i>	<i>FREM2</i>	<i>IFT57</i>	<i>KIAA0753</i>	<i>PAX2</i>	<i>SEC61A1</i>	<i>TCTN1</i>	<i>WDPCP</i>	
<i>ARMC9</i>	<i>CC2D2A</i>	<i>DDX59</i>	<i>GANAB</i>	<i>IFT74</i>	<i>KIF7</i>	<i>PAX8</i>	<i>SIX1</i>	<i>TMEM67</i>	<i>WDR19</i>	
<i>ATXN10</i>	<i>CCDC28B</i>	<i>DSTYK</i>	<i>GATA3</i>	<i>IFT80</i>	<i>KIF14</i>	<i>PBX1</i>	<i>SIX2</i>	<i>TMEM107</i>	<i>WDR34</i>	
<i>B9D1</i>	<i>CENPF</i>	<i>DYNC2H1</i>	<i>GDNF</i>	<i>IFT81</i>	<i>LMX1B</i>	<i>PDE6D</i>	<i>SIX5</i>	<i>TMEM138</i>	<i>WDR35</i>	
<i>B9D2</i>	<i>CEP41</i>	<i>DZIP1L</i>	<i>GLIS2</i>	<i>IFT122</i>	<i>LRP5</i>	<i>PIBF1</i>	<i>SLIT2</i>	<i>TMEM216</i>	<i>WDR60</i>	

Supplementary Table S5. The gene list constructed using HaloPlex (version 7, 181 genes)

<i>ACE</i>	<i>BBS1</i>	<i>CEP83</i>	<i>EP300</i>	<i>GFRA1</i>	<i>IFT80</i>	<i>LRIG2</i>	<i>PIBF1</i>	<i>SOX11</i>	<i>TNXB</i>	<i>ZNF423</i>
<i>ACTG2</i>	<i>BBS2</i>	<i>CEP104</i>	<i>EVC</i>	<i>GLIS2</i>	<i>IFT81</i>	<i>LRP5</i>	<i>PKD1</i>	<i>SOX17</i>	<i>TRAF3IP1</i>	
<i>AGT</i>	<i>BBS4</i>	<i>CEP120</i>	<i>EVC2</i>	<i>GLIS3</i>	<i>IFT122</i>	<i>LZTFL1</i>	<i>PKD2</i>	<i>SPRY1</i>	<i>TRIM32</i>	
<i>AGTR1</i>	<i>BBS5</i>	<i>CEP164</i>	<i>EXOC4</i>	<i>GPC3</i>	<i>IFT140</i>	<i>MAPKBP1</i>	<i>PKHD1</i>	<i>SRGAP1</i>	<i>TSC1</i>	
<i>AGTR2</i>	<i>BBS7</i>	<i>CEP290</i>	<i>EXOC8</i>	<i>GREB1L</i>	<i>IFT172</i>	<i>MKKS</i>	<i>REN</i>	<i>SUFU</i>	<i>TSC2</i>	
<i>AHI1</i>	<i>BBS9</i>	<i>CHD1L</i>	<i>EYA1</i>	<i>GREM1</i>	<i>INPP5E</i>	<i>MKS1</i>	<i>RET</i>	<i>TBC1D32</i>	<i>TTC8</i>	
<i>ALG8</i>	<i>BBS10</i>	<i>CHD4</i>	<i>FAN1</i>	<i>GRIP1</i>	<i>INTU</i>	<i>MUC1</i>	<i>ROBO2</i>	<i>TBX1</i>	<i>TTC21B</i>	
<i>ALG9</i>	<i>BBS12</i>	<i>CHD7</i>	<i>FGF20</i>	<i>HNF1B</i>	<i>INVS</i>	<i>NEK1</i>	<i>RPGRIP1L</i>	<i>TBX18</i>	<i>UMOD</i>	
<i>ALMS1</i>	<i>BICC1</i>	<i>CHRM3</i>	<i>FGFR1</i>	<i>HOXA13</i>	<i>IQCB1</i>	<i>NEK8</i>	<i>SALL1</i>	<i>TCTEX1D2</i>	<i>UPK3A</i>	
<i>ANKS6</i>	<i>C2CD3</i>	<i>CITED1</i>	<i>FGFR2</i>	<i>HPRT1</i>	<i>ITGA8</i>	<i>NOTCH2</i>	<i>SARS2</i>	<i>TCTN1</i>	<i>VANGL2</i>	
<i>ARL3</i>	<i>C5orf42</i>	<i>CRB2</i>	<i>FRAS1</i>	<i>HPSE2</i>	<i>JAG1</i>	<i>NPHP1</i>	<i>SCLT1</i>	<i>TCTN2</i>	<i>WDPCP</i>	
<i>ARL6</i>	<i>C8orf37</i>	<i>CSPP1</i>	<i>FREM1</i>	<i>HYLS1</i>	<i>KAL1</i>	<i>NPHP3</i>	<i>SDCCAG8</i>	<i>TCTN3</i>	<i>WDR19</i>	
<i>ARL13B</i>	<i>CC2D2A</i>	<i>DCDC2</i>	<i>FREM2</i>	<i>INF2</i>	<i>KIAA0556</i>	<i>NPHP4</i>	<i>SEC61A1</i>	<i>TMEM67</i>	<i>WDR34</i>	
<i>ARMC9</i>	<i>CCDC28B</i>	<i>DDX59</i>	<i>GANAB</i>	<i>IFT27</i>	<i>KIAA0586</i>	<i>OFD1</i>	<i>SIX1</i>	<i>TMEM107</i>	<i>WDR35</i>	
<i>ATXN10</i>	<i>CDC5L</i>	<i>DNAJB11</i>	<i>GATA3</i>	<i>IFT43</i>	<i>KIAA0753</i>	<i>PAX2</i>	<i>SIX2</i>	<i>TMEM138</i>	<i>WDR60</i>	
<i>B9D1</i>	<i>CDKN1C</i>	<i>DSTYK</i>	<i>GDF11</i>	<i>IFT52</i>	<i>KIF7</i>	<i>PAX8</i>	<i>SIX5</i>	<i>TMEM216</i>	<i>WNT4</i>	
<i>B9D2</i>	<i>CENPF</i>	<i>DYNC2H1</i>	<i>GNDF</i>	<i>IFT57</i>	<i>KIF14</i>	<i>PBX1</i>	<i>SLIT2</i>	<i>TMEM231</i>	<i>WT1</i>	
<i>BBIP1</i>	<i>CEP41</i>	<i>DZIP1L</i>	<i>GEN1</i>	<i>IFT74</i>	<i>LMX1B</i>	<i>PDE6D</i>	<i>SOX9</i>	<i>TMEM237</i>	<i>XPNPEP3</i>	

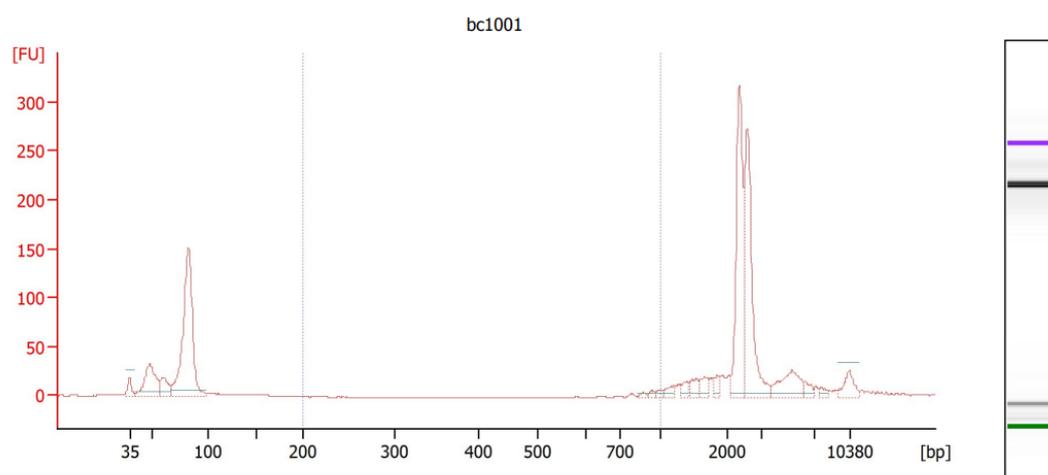
Supplementary Table S6. The gene list constructed using HaloPlex (version 8, 183 genes)

<i>ACE</i>	<i>BBS2</i>	<i>CEP164</i>	<i>EYA1</i>	<i>HNF1B</i>	<i>ITGA8</i>	<i>NPHP3</i>	<i>SIX1</i>	<i>TMEM138</i>	<i>WT1</i>
<i>ACTG2</i>	<i>BBS4</i>	<i>CEP290</i>	<i>FAN1</i>	<i>HOXA13</i>	<i>JAG1</i>	<i>NPHP4</i>	<i>SIX2</i>	<i>TMEM216</i>	<i>XPNPEP3</i>
<i>ADAMTS9</i>	<i>BBS5</i>	<i>CHD1L</i>	<i>FGF20</i>	<i>HPRT1</i>	<i>KAL1</i>	<i>OFD1</i>	<i>SIX5</i>	<i>TMEM231</i>	<i>ZNF423</i>
<i>AGT</i>	<i>BBS7</i>	<i>CHD4</i>	<i>FGFR1</i>	<i>HPSE2</i>	<i>KIAA0556</i>	<i>PAX2</i>	<i>SLIT2</i>	<i>TMEM237</i>	
<i>AGTR1</i>	<i>BBS9</i>	<i>CHD7</i>	<i>FGFR2</i>	<i>HYLS1</i>	<i>KIAA0586</i>	<i>PAX8</i>	<i>SON</i>	<i>TNXB</i>	
<i>AGTR2</i>	<i>BBS10</i>	<i>CHRM3</i>	<i>FRAS1</i>	<i>INF2</i>	<i>KIAA0753</i>	<i>PBX1</i>	<i>SOX9</i>	<i>TRAF3IP1</i>	
<i>AHI1</i>	<i>BBS12</i>	<i>CITED1</i>	<i>FREM1</i>	<i>IFT27</i>	<i>KIF7</i>	<i>PDE6D</i>	<i>SOX11</i>	<i>TRIM32</i>	
<i>ALG8</i>	<i>BICC1</i>	<i>CRB2</i>	<i>FREM2</i>	<i>IFT43</i>	<i>KIF14</i>	<i>PIBF1</i>	<i>SOX17</i>	<i>TSC1</i>	
<i>ALG9</i>	<i>C2CD3</i>	<i>CSPP1</i>	<i>GANAB</i>	<i>IFT52</i>	<i>LMX1B</i>	<i>PKD1</i>	<i>SPRY1</i>	<i>TSC2</i>	
<i>ALMS1</i>	<i>C5orf42</i>	<i>DCDC2</i>	<i>GATA3</i>	<i>IFT57</i>	<i>LRIG2</i>	<i>PKD2</i>	<i>SRGAP1</i>	<i>TTC8</i>	
<i>ANKS6</i>	<i>C8orf37</i>	<i>DDX59</i>	<i>GDF11</i>	<i>IFT74</i>	<i>LRP5</i>	<i>PKHD1</i>	<i>SUFU</i>	<i>TTC21B</i>	
<i>ARL3</i>	<i>CC2D2A</i>	<i>DNAJB11</i>	<i>GDNF</i>	<i>IFT80</i>	<i>LZTFL1</i>	<i>REN</i>	<i>TBC1D32</i>	<i>UMOD</i>	
<i>ARL6</i>	<i>CCDC28B</i>	<i>DSTYK</i>	<i>GEN1</i>	<i>IFT81</i>	<i>MAPKBP1</i>	<i>RET</i>	<i>TBX1</i>	<i>UPK3A</i>	
<i>ARL13B</i>	<i>CDC5L</i>	<i>DYNC2H1</i>	<i>GFRA1</i>	<i>IFT122</i>	<i>MKKS</i>	<i>ROBO2</i>	<i>TBX18</i>	<i>VANGL2</i>	
<i>ARMC9</i>	<i>CDKN1C</i>	<i>DZIP1L</i>	<i>GLIS2</i>	<i>IFT140</i>	<i>MKS1</i>	<i>RPGRIP1L</i>	<i>TCTEX1D2</i>	<i>WDPCP</i>	
<i>ATXN10</i>	<i>CENPF</i>	<i>EP300</i>	<i>GLIS3</i>	<i>IFT172</i>	<i>MUC1</i>	<i>SALL1</i>	<i>TCTN1</i>	<i>WDR19</i>	
<i>B9D1</i>	<i>CEP41</i>	<i>EVC</i>	<i>GPC3</i>	<i>INPP5E</i>	<i>NEK1</i>	<i>SARS2</i>	<i>TCTN2</i>	<i>WDR34</i>	
<i>B9D2</i>	<i>CEP83</i>	<i>EVC2</i>	<i>GREB1L</i>	<i>INTU</i>	<i>NEK8</i>	<i>SCLT1</i>	<i>TCTN3</i>	<i>WDR35</i>	
<i>BBIP1</i>	<i>CEP104</i>	<i>EXOC4</i>	<i>GREM1</i>	<i>INVS</i>	<i>NOTCH2</i>	<i>SDCCAG8</i>	<i>TMEM67</i>	<i>WDR60</i>	
<i>BBS1</i>	<i>CEP120</i>	<i>EXOC8</i>	<i>GRIP1</i>	<i>IQCB1</i>	<i>NPHP1</i>	<i>SEC61A1</i>	<i>TMEM107</i>	<i>WNT4</i>	

Supplementary Table S8. The identified sequences of VNTR region for each patient analyzed by LRS

SC270 / bc1001

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
bc1001-0	2370	3368	0.337	Correct	-	30
bc1001-1	2670	2977	0.298	Correct	-	35
bc1001-2	2371	303	0.030	Error	/	/
bc1001-3	2371	259	0.026	Error	/	/



Peak table for sample 1 : **bc1001**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	18.3	Lower Marker	0.9	8.9
2	48	537.83	16,908.4	32.4		2.5	46.1
3	60	196.96	4,999.5	19.1		1.0	17.6
4	82	1,855.11	34,317.2	150.4		3.4	174.0
5	870	13.05	22.7	5.7		1.0	4.0
6	931	15.85	25.8	7.4		0.8	5.0
7	1,006	18.89	28.5	8.4		0.7	6.2
8	1,188	34.31	43.8	12.7		1.0	11.7
9	1,384	37.02	40.5	16.5		0.8	13.1
10	1,524	49.92	49.6	19.6		0.9	18.2
11	1,650	53.05	48.7	21.3		0.9	19.8
12	1,840	36.18	29.8	22.9		0.6	14.0
13	2,333	635.82	412.9	319.5		1.3	255.0
14	2,582	646.18	379.2	274.4		2.6	261.1
15	5,248	163.35	47.2	27.6		3.1	67.6
16	6,236	33.48	8.1	16.4		1.0	13.9
17	7,614	18.97	3.8	10.2		0.9	8.1
18	10,380	75.00	10.9	27.6	Upper Marker	2.2	34.5

bc1001-0

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bc1001-1

GGAGAA

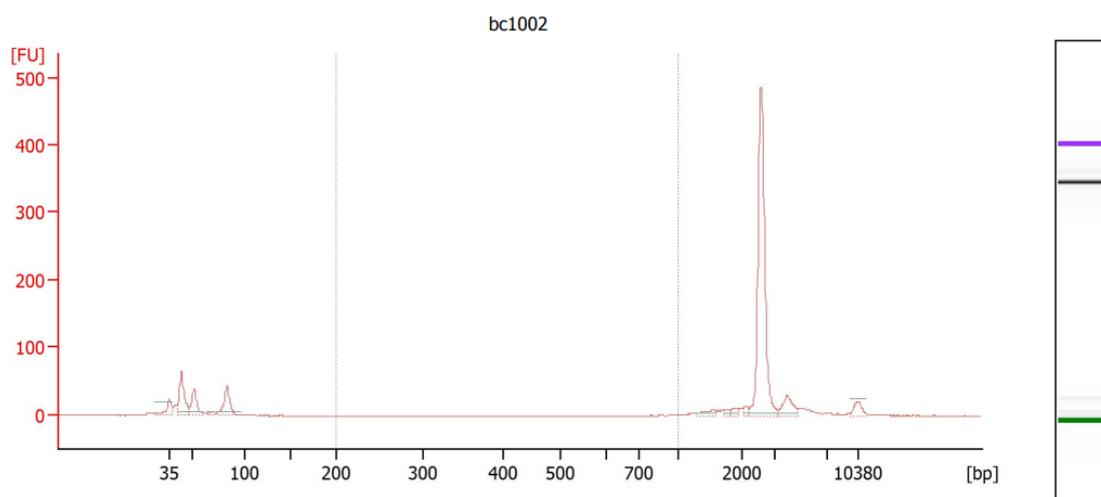
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SC298 / bc1002

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1002-0	2610	3097	0.310	Correct	-	34
1002-1	2670	2926	0.293	Correct	-	35
1002-2	2611	451	0.045	Error		
1002-3	2654	229	0.023	Error		



Peak table for sample 1 : bc1002

Peak	Size [bp]	Conc. [pg/ μ l]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	24.4	Lower Marker	1.8	18.7
2	43	818.65	28,838.2	65.1		1.3	45.3
3	52	480.19	13,928.9	39.5		1.5	28.5
4	70	67.26	1,458.6	5.2		0.8	4.2
5	83	569.99	10,354.8	44.3		2.5	36.4
6	1,548	65.03	63.7	9.0		2.0	16.2
7	1,793	28.19	23.8	10.7		0.7	7.3
8	1,895	36.32	29.0	11.8		0.8	9.6
9	2,101	32.31	23.3	13.8		0.6	8.8
10	2,593	1,623.49	948.8	487.7		3.0	447.2
11	3,985	150.80	57.3	31.0		2.0	42.3
12	10,380	75.00	10.9	21.6	Upper Marker	1.8	23.5

bc1002-1

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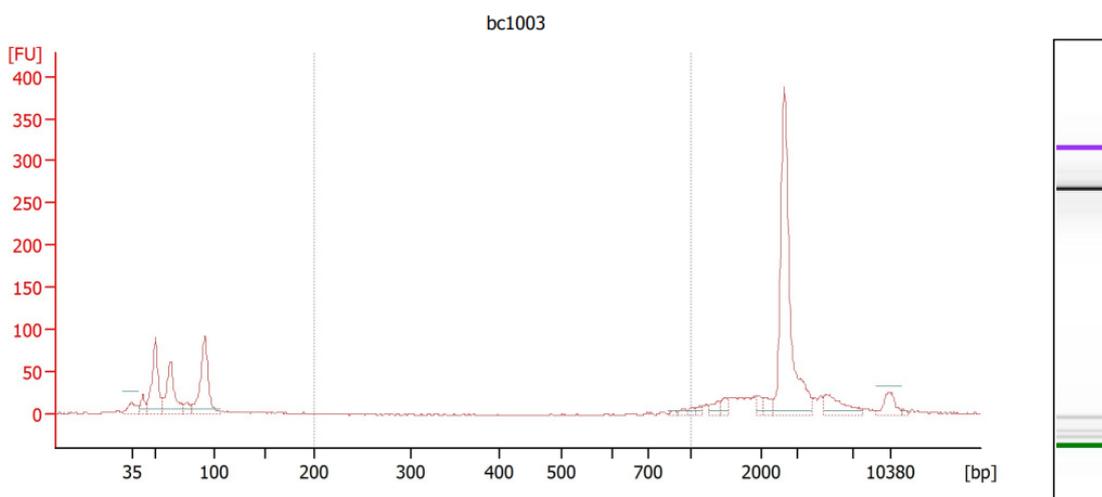
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SC356 / bc1003

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
1003-0	2731	2895	0.290	Correct	10 / C	36
1003-1	2670	1005	0.101	Correct	-	35
1003-2	2723	435	0.044	Error		
1003-3	2671	257	0.026	Error		



Peak table for sample 2 : bc1003

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	14.2	Lower Marker	1.6	14.4
2	42	136.04	4,895.1	22.7		0.7	11.7
3	50	701.65	21,332.7	90.1		1.4	65.0
4	62	645.24	15,657.0	62.4		2.1	61.7
5	76	84.77	1,681.8	13.3		0.6	8.4
6	92	847.25	14,018.5	92.2		2.7	86.6
7	883	14.79	25.4	6.6		1.0	4.9
8	952	18.46	29.4	8.1		0.9	6.4
9	1,014	18.46	27.6	10.0		0.7	6.6
10	1,113	18.85	25.7	11.6		0.6	6.8
11	1,374	40.81	45.0	17.4		1.0	15.6
12	1,480	42.63	43.7	21.7		0.8	16.6
13	1,952	33.87	26.3	22.8		0.6	14.4
14	2,087	49.39	35.9	21.8		1.0	21.3
15	2,630	985.41	567.8	391.3		3.7	431.1
16	5,039	133.45	40.1	24.7		3.7	59.7
17	10,380	75.00	10.9	27.2	Upper Marker	2.4	37.4

bc1003-0

GGAGAA

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bc1003-1

GGAGAA

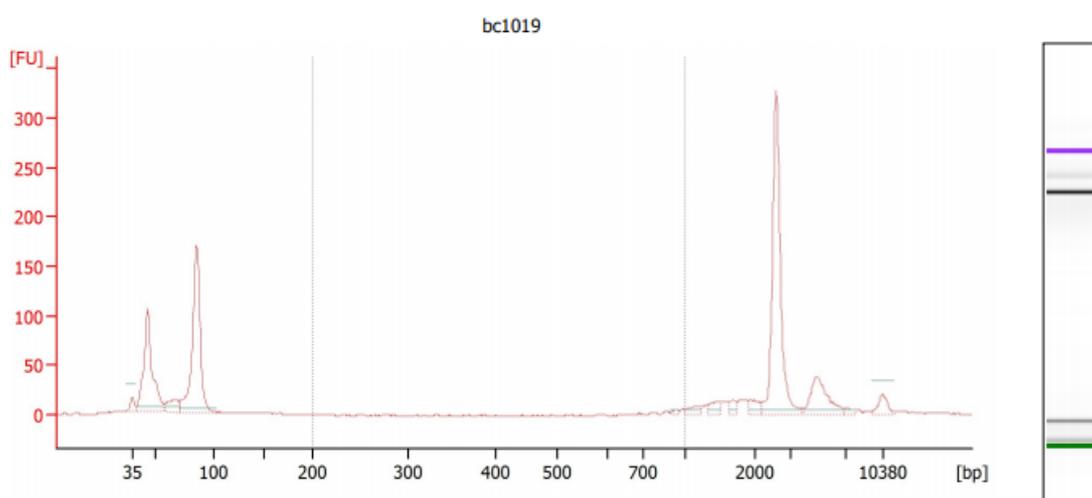
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SC360 / bc1019

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1019-0	2610	3214	0.321	Correct	-	34
1019-1	2670	3036	0.304	Correct	-	35
1019-2	2671	175	0.018	Error		
1019-3	895	172	0.017	Error		



Peak table for sample 8 : bc1019

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	14.5	Lower Marker	0.9	6.4
2	45	1,833.26	62,373.2	103.9		2.6	91.1
3	67	272.90	6,159.4	12.6		1.4	14.8
4	85	2,399.24	42,778.6	169.1		3.4	135.0
5	930	17.74	28.9	5.5		0.8	3.2
6	1,171	54.80	70.9	9.0		1.5	10.8
7	1,437	65.18	68.7	13.4		1.2	13.5
8	1,671	42.07	38.2	14.9		0.7	9.1
9	1,921	63.74	50.3	15.4		1.1	14.4
10	2,575	1,198.74	705.3	328.4		3.7	279.1
11	4,797	283.19	89.4	37.6		3.8	67.3
12	6,908	21.71	4.8	6.5		0.9	5.2
13	10,380	75.00	10.9	20.4	Upper Marker	2.2	19.8

bc1019-1 (complementary sequence)

GGAGAA

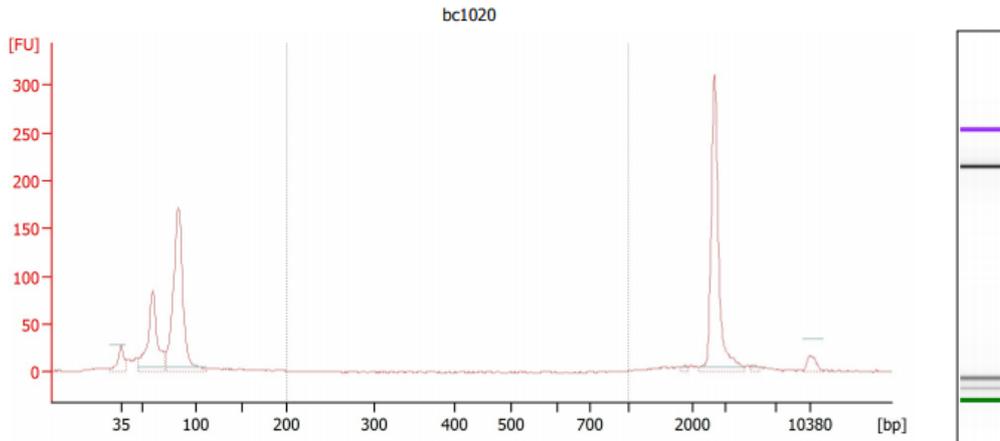
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SC362 / bc1020

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1020-0	2730	3867	0.387	Correct	-	36
1020-1	2670	3578	0.358	Correct	-	35
1020-2	2671	618	0.062	Error		
1020-3	2671	204	0.020	Error		



Peak table for sample 9 : bc1020

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	27.0	Lower Marker	1.7	19.5
2	59	1,777.90	45,573.6	85.5		2.7	88.2
3	83	3,671.20	67,109.4	171.7		4.2	191.3
4	1,885	20.12	16.2	6.7		0.7	4.2
5	2,653	1,239.28	707.7	312.3		4.6	268.1
6	5,054	22.00	6.6	6.8		0.8	4.9
7	10,380	75.00	10.9	17.2	Upper Marker	2.2	18.4

bc1020-0

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bc1020-1

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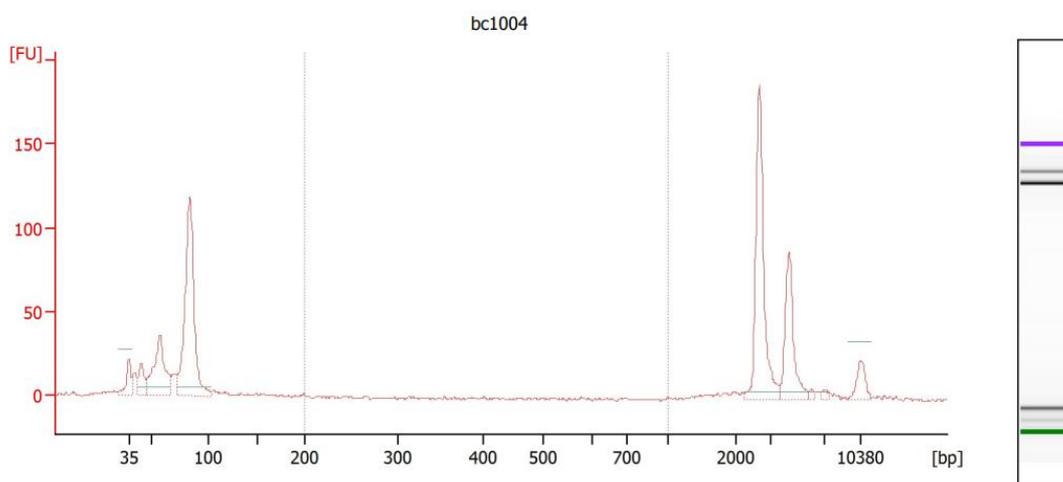
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SC370 / bc1004

Sequence ID	Bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
bc1004-0	2731	3250	0.392	Correct	5 /G	36
bc1004-1	4290	985	0.119	Correct	-	62
bc1004-2	2732	299	0.036	Error		
bc1004-3	2732	259	0.031	Error		



Peak table for sample 3 : **bc1004**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	21.7	Lower Marker	1.4	12.7
2	43	210.80	7,420.5	18.9		0.9	12.7
3	57	706.66	18,719.3	36.0		2.3	46.1
4	83	1,820.28	33,177.5	119.2		3.3	125.6
5	2,668	590.46	335.3	188.4		3.3	173.3
6	4,335	293.24	102.5	88.3		2.8	87.5
7	5,962	13.50	3.4	6.8		0.7	4.1
8	6,998	13.18	2.9	5.8		0.7	4.0
9	10,380	75.00	10.9	23.8	Upper Marker	2.3	25.0

bc1004-0

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bc1004-1

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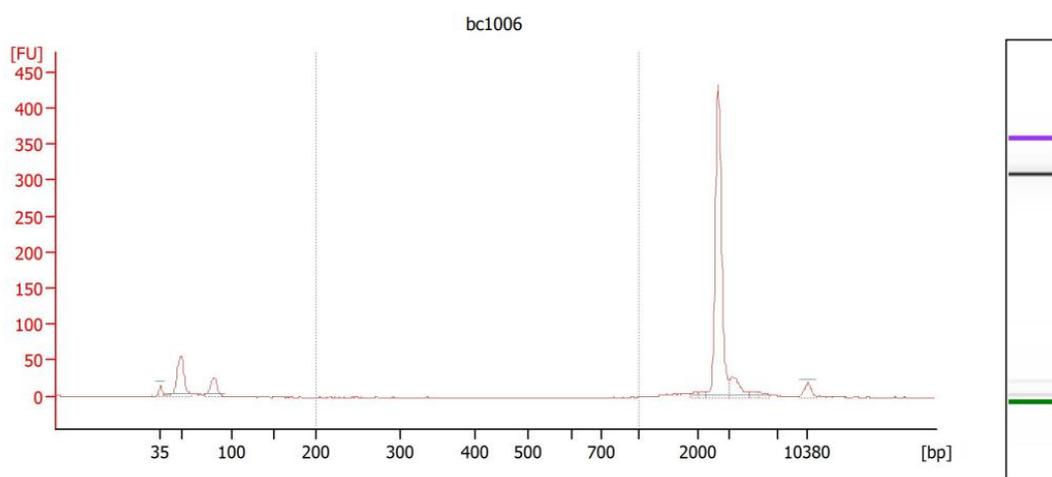
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GCTTCTACTCTGGTGACAACGGC

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SC 416 / bc1006

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
bc1006-0	2670	3889	0.389	Correct	-	35
bc1006-1	2611	3095	0.310	Correct	6/C	34
bc1006-2	2671	179	0.018	Error		
bc1006-3	2614	107	0.011	Error		



Peak table for sample 2 : bc1006

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	16.0	Lower Marker	1.0	8.1
2	50	1,072.74	32,474.7	56.4		2.9	61.1
3	83	461.59	8,437.9	27.2		2.3	28.3
4	1,938	32.06	25.1	9.1		0.9	8.2
5	2,122	27.50	19.6	8.8		0.8	7.2
6	2,634	1,471.61	846.7	436.2		2.5	388.9
7	3,230	174.02	81.6	30.3		2.2	46.6
8	4,969	32.62	9.9	9.3		0.9	8.8
9	5,480	36.63	10.1	8.9		1.3	9.9
10	10,380	75.00	10.9	21.2	Upper Marker	2.0	22.5

bc1006-0 (complementary sequence)

GGAGAA

AAGGAGACTTCGGCTACCCAGAGAAGTTTCAGTGCCAGCTCTACTGAGAAGAATGCTGTG
AGTATGACCAGCAGCGTACTCTCCAGCCACAGCCCGGTTTCAGGCTCCTCCACCACTCAG
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GCCCACGGTGTACCTCGGCCCCGGACACCAGCCGGCCCCGGGTCCACCGCCCCCAA
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GGCTCCACCGCCCCCTCAGTCCACAATGTCACCTCGGCTCAGGCTCTGCATCAGGCTCA
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bc1006-1 (complementary sequence)

GGAGAA

AAGGAGACTTCGGCTACCCAGAGAAGTTTCAGTGCCAGCTCTACTGAGAAGAATGCTGTG
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GGACAGGATGTCACCTCGCCCCGGCCACGGAACAGCTTCAGGTTTCAGCTGCCACCTGG
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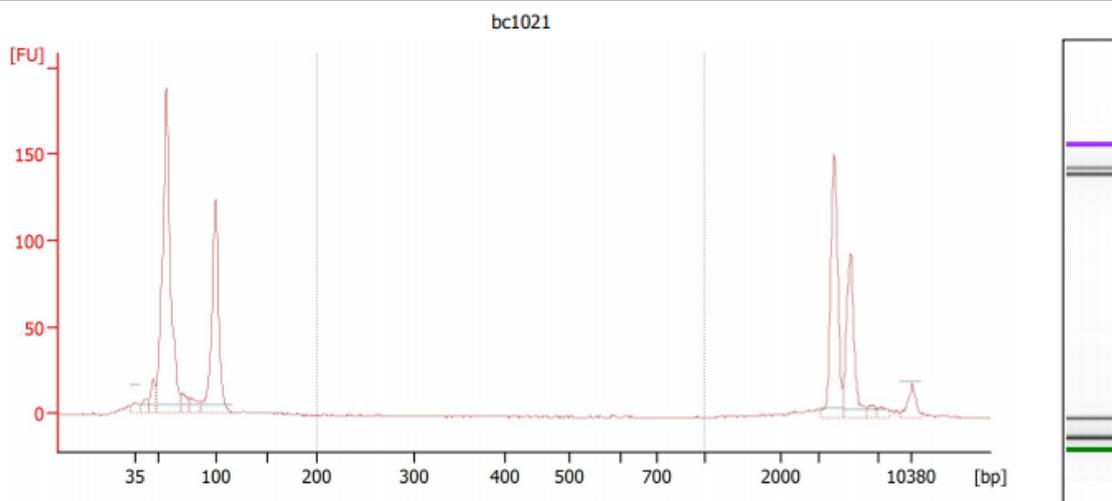
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GTTTCTACTCTGGTGCACAACGGC

SC422 / bc1021

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1021-0	3870	3967	0.397	Correct	-	55
1021-1	4950	1335	0.134	Correct	-	73
1021-2	2143	140	0.014	Error		
1021-3	3871	132	0.013	Error		



Peak table for sample 2 : bc1021

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	6.1	Lower Marker	0.8	4.8
2	41	129.93	4,748.1	7.9		0.7	5.5
3	46	238.65	7,795.3	20.4		0.6	10.6
4	56	3,706.63	99,879.3	188.7		2.2	172.0
5	71	160.68	3,446.0	11.7		0.7	7.7
6	77	164.87	3,254.0	8.7		1.0	8.0
7	99	1,958.85	29,961.6	123.6		2.8	99.7
8	3,990	588.08	223.3	152.9		2.0	128.5
9	5,114	385.13	114.1	95.1		2.1	84.7
10	6,409	27.52	6.5	7.2		0.9	6.1
11	7,185	30.88	6.5	6.0		1.2	6.9
12	10,380	75.00	10.9	20.2	Upper Marker	1.9	18.3

bc1021-0 (complementary sequence)

GGAGAA

AAGGAGACTTCGGCTACCCAGAGAAGTTCAGTGCCCAGCTCTACTGAGAAGAATGCTGTG
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bc1021-1 (complementary sequence)

GGAGAA

AAGGAGACTTCGGCTACCCAGAGAAGTTCAGTGCCAGCTCTACTGAGAAGAATGCTGTG
AGTATGACCAGCAGCGTACTCTCCAGCCACAGCCCGGTTCAAGGCTCCTCCACCACTCAG
GGACAGGATGTCACCTCGGCCCCGGCCACGGAACCAGCTTCAGGTTCACTGCCACCTGG
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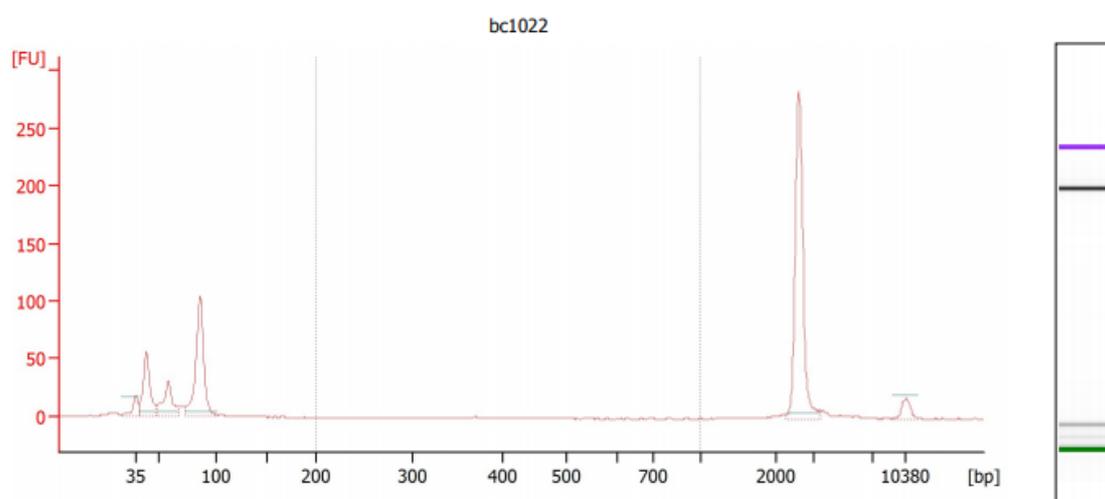
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GCTTCTACTCTGGTGCACAACGGC	

SC431 / bc1022

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1022-0	2670	3235	0.324	Correct	-	35
1022-1	2610	2812	0.281	Correct	-	34
1022-2	2611	652	0.065	Error		
1022-3	2671	316	0.032	Error		



Peak table for sample 3 : **bc1022**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	18.6	Lower Marker	1.7	12.3
2	42	953.97	34,574.5	56.5		1.5	42.8
3	58	632.73	16,616.9	30.7		1.9	31.1
4	86	1,764.93	31,178.2	105.2		3.0	91.7
5	2,603	1,135.22	660.8	285.4		3.1	246.6
6	10,380	75.00	10.9	18.3	Upper Marker	2.3	18.5

bc1022-1

GGAGAA

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AGTATGACCAGCAGCGTACTCTCCAGCCACAGCCCGGTTTCAGGCTCTCCACCACTCAG
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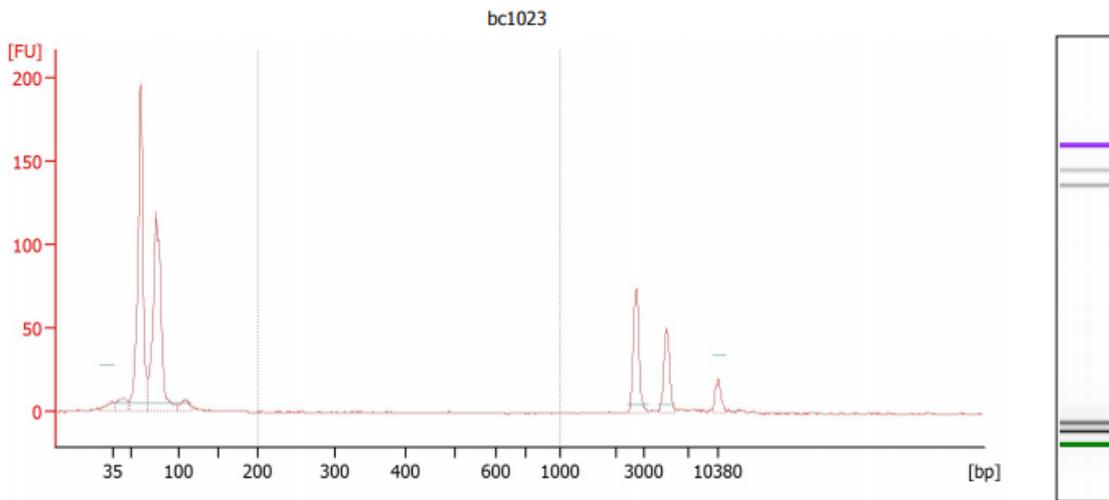
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SC449 / bc1023

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1023-0	2670	3789	0.379	Correct	-	35
1023-1	2671	642	0.064	Error		
1023-2	2670	621	0.062	Error		
1023-3	4770	548	0.055	Correct	-	70



Peak table for sample 10 : bc1023

Peak	Size [bp]	Conc. [pg/ μ l]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	5.6	Lower Marker	1.7	6.4
2	44	280.28	9,759.2	8.3		1.6	10.7
3	59	3,232.49	82,613.9	196.2		2.2	134.6
4	76	2,986.94	59,888.6	119.3		3.3	129.2
5	109	172.97	2,414.7	7.1		1.6	8.1
6	2,728	301.27	167.3	74.9		2.5	58.0
7	5,006	203.60	61.6	51.3		1.9	39.9
8	10,380	75.00	10.9	20.6	Upper Marker	1.6	16.4

bc1023-0

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bc1023-3

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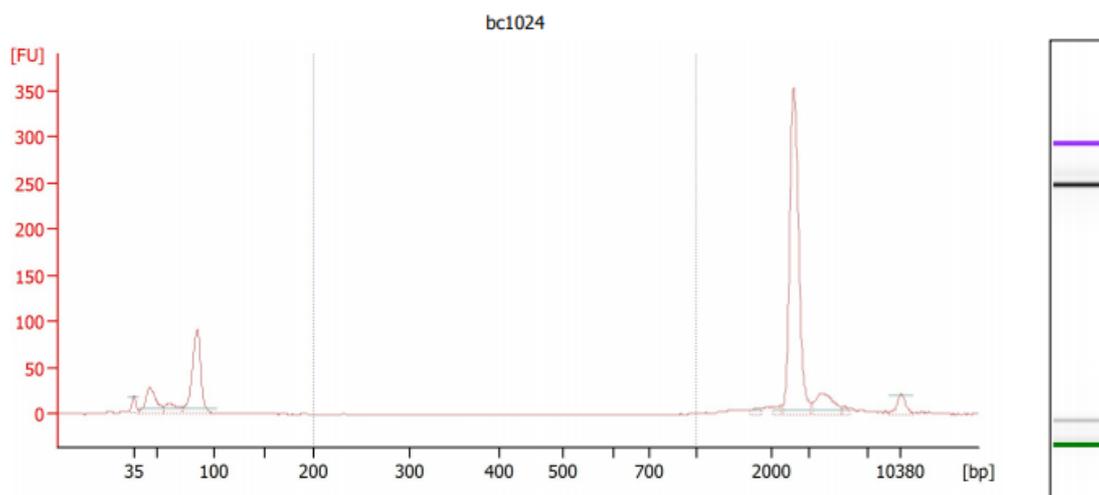
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SC465 / bc1024

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1024-0	2550	3263	0.326	Correct	-	33
1024-1	2670	2648	0.265	Correct	-	35
1024-2	2671	273	0.027	Error		
1024-3	2669	247	0.025	Error		



Peak table for sample 4 : **bc1024**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	17.7	Lower Marker	1.0	7.8
2	45	568.15	19,254.0	26.9		2.3	31.4
3	60	225.49	5,686.7	9.5		1.6	13.4
4	85	1,366.36	24,411.7	90.5		3.2	85.1
5	1,867	29.30	23.8	7.2		1.1	7.3
6	2,259	31.22	20.9	9.7		0.9	8.0
7	2,596	1,338.49	781.3	355.7		2.6	347.1
8	3,933	175.96	67.8	23.2		2.7	46.4
9	5,378	23.26	6.6	9.6		0.7	6.2
10	10,380	75.00	10.9	22.8	Upper Marker	2.2	22.1

bc1024-0

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bc1024-1

GGAGAA

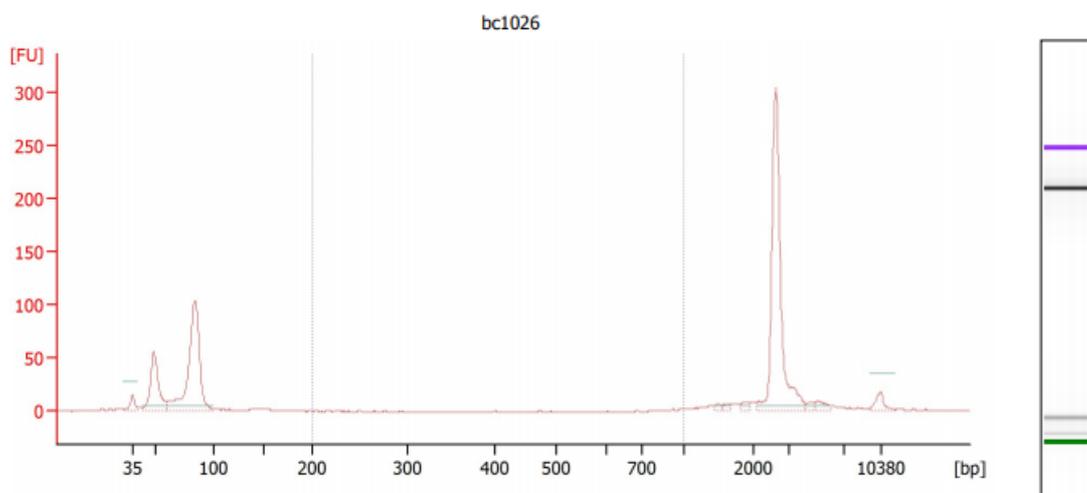
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SC469 / bc1026

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1026-0	2670	3339	0.334	Correct	-	35
1026-1	2730	3253	0.325	Correct	-	36
1026-2	2731	527	0.053	Error		
1026-3	2246	173	0.017	Error		



Peak table for sample 11 : bc1026

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	14.6	Lower Marker	1.4	7.7
2	49	1,137.21	35,315.1	56.3		2.2	45.6
3	83	2,684.03	48,920.8	103.4		4.3	116.8
4	1,490	24.52	24.9	7.1		0.7	4.0
5	1,611	27.32	25.7	6.8		0.7	4.5
6	1,861	33.69	27.4	8.2		0.8	5.8
7	2,618	1,543.75	893.3	304.7		4.6	278.5
8	4,709	32.82	10.6	8.1		0.8	6.0
9	5,075	54.71	16.3	9.1		1.5	10.1
10	10,380	75.00	10.9	18.2	Upper Marker	2.4	15.4

bc1026-0

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bc1025-1

GGAGAA

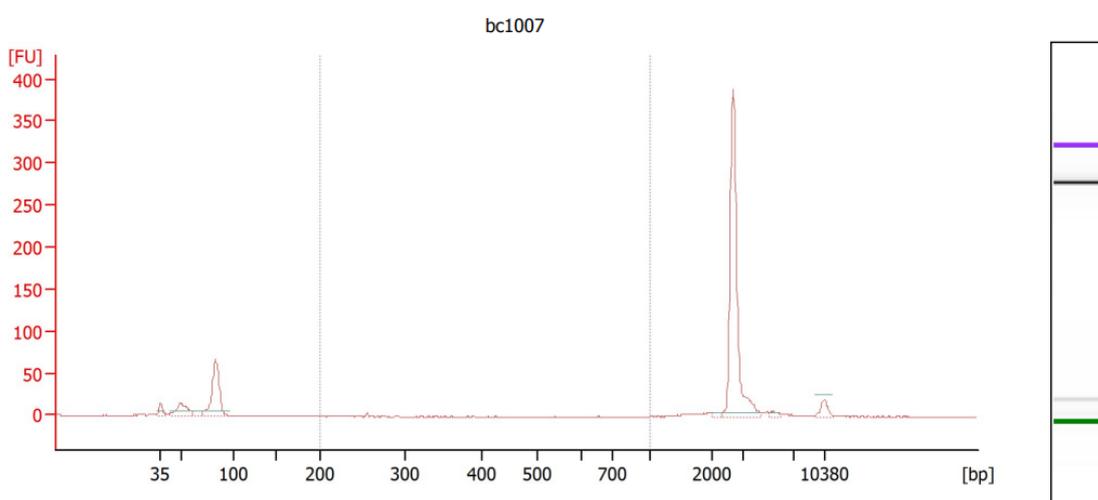
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SC489 / bc1007

Sequence ID	Bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
bc1007-0	2670	3146	0.315	Correct	-	35
bc1007-1	2731	1785	0.179	Correct	7/C	36
bc1007-2	2671	411	0.041	Error		
bc1007-3	2732	379	0.038	Error		



Peak table for sample 3 : bc1007

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	15.4	Lower Marker	1.0	7.5
2	49	440.26	13,579.4	15.6		2.5	22.9
3	64	102.13	2,431.4	6.1		0.9	5.5
4	83	1,162.81	21,294.4	67.2		2.8	65.5
5	2,188	27.61	19.1	6.4		1.1	6.6
6	2,655	1,499.93	856.0	390.8		4.1	363.2
7	5,153	29.65	8.7	6.7		1.2	7.3
8	10,380	75.00	10.9	20.3	Upper Marker	2.0	20.6

bc1007-0

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bc1007-1

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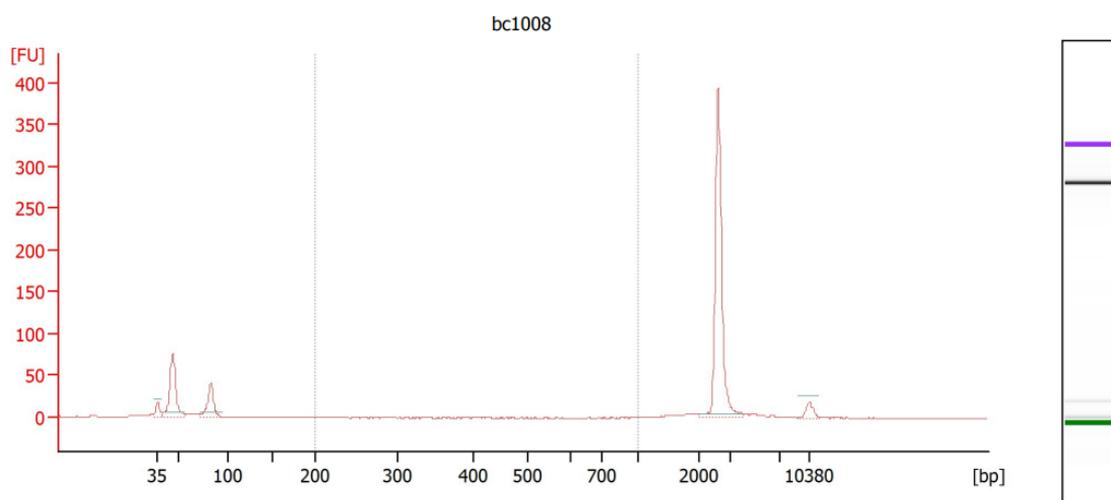
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repeat 32
repeat 33
repeat 34
repeat 35
repeat 36

SC507 / bc1008

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1008-0	2670	3390	0.339	Correct	-	35
1008-1	2670	2567	0.257	Correct	-	35
1008-2	2671	429	0.043	Error		
1008-3	2671	314	0.031	Error		



Peak table for sample 4 : bc1008

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	17.6	Lower Marker	0.8	8.6
2	46	1,338.45	44,052.0	75.6		2.5	62.1
3	82	614.45	11,293.9	40.7		2.5	31.8
4	2,606	1,554.25	903.8	395.5		4.8	343.6
5	10,380	75.00	10.9	19.5	Upper Marker	2.2	18.9

bc1008-0

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bc1008-1

GGAGAA

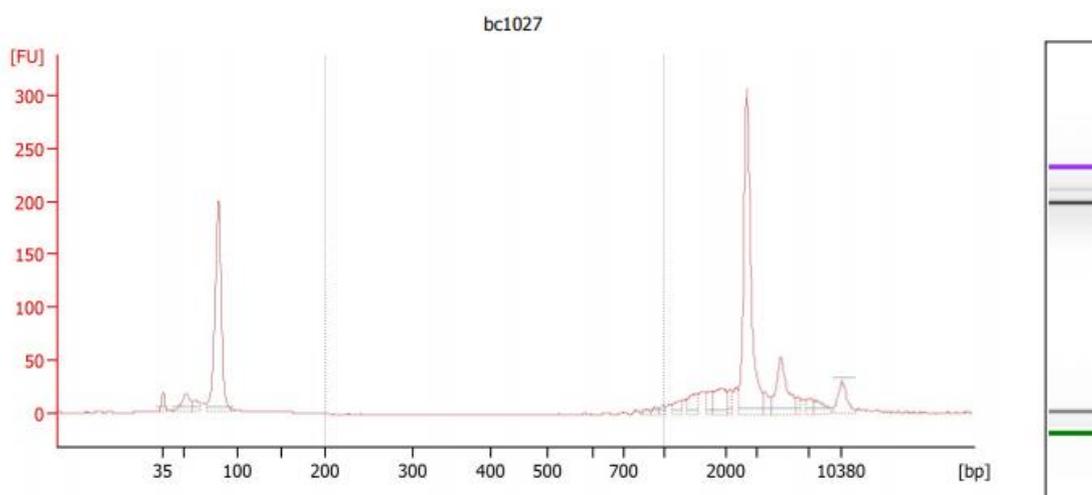
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SC511 / bc1027

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1027-0	2730	1889	0.189	Correct	-	36
1027-1	2731	257	0.026	Error		
1027-2	2731	242	0.024	Error		
1027-3	4830	163	0.016	Correct	-	71



Peak table for sample 1 : **bc1027**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	18.4	Lower Marker	1.2	8.2
2	52	204.73	6,008.7	17.4		1.9	19.5
3	60	99.91	2,529.6	10.9		0.9	9.7
4	82	1,716.61	31,547.1	200.6		2.9	175.6
5	872	12.90	22.4	5.5		1.0	4.3
6	935	14.27	23.1	7.3		0.8	5.0
7	997	16.17	24.6	8.9		0.7	5.8
8	1,278	37.02	43.9	14.0		1.1	14.1
9	1,542	61.98	60.9	20.1		1.2	24.8
10	1,736	33.40	29.2	22.4		0.6	13.9
11	1,916	86.51	68.4	26.1		1.4	37.1
12	2,067	36.37	26.7	23.3		0.6	15.9
13	2,660	687.33	391.6	308.6		2.5	305.4
14	3,477	37.39	16.3	21.2		0.8	16.8
15	4,814	172.25	54.2	54.5		2.5	78.1
16	5,983	23.57	6.0	16.7		0.6	10.8
17	6,886	28.53	6.3	14.9		0.8	13.1
18	7,661	40.04	7.9	13.2		1.8	18.7
19	10,380	75.00	10.9	31.0	Upper Marker	2.4	38.0

bc1027-3

GGAGAA

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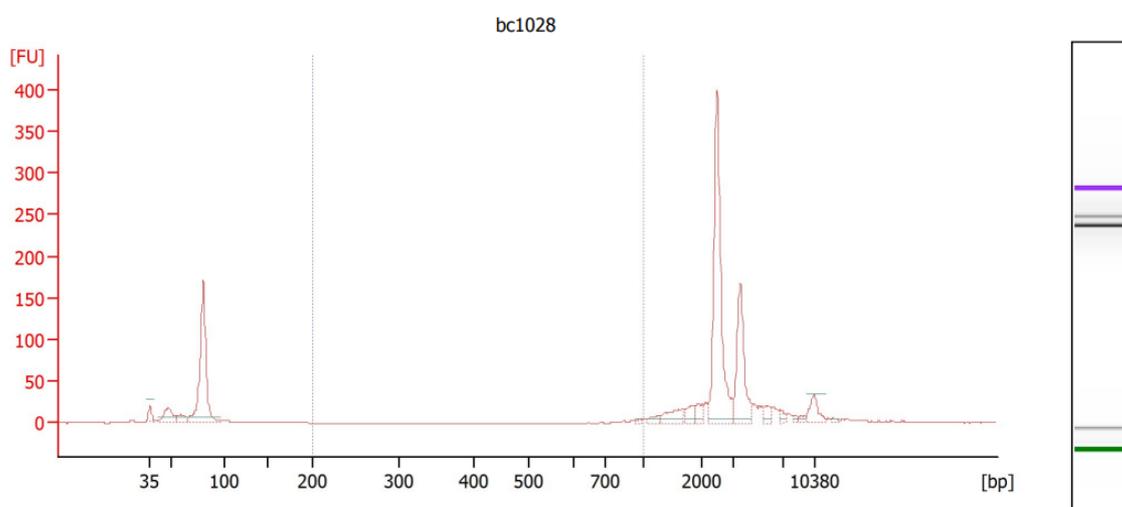
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SC512 / bc1028

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
1028-0	2610	3816	0.382	Correct	-	34
1028-1	3871	874	0.087	Correct	14/C	55
1028-2	2611	457	0.046	Error		
1028-3	2099	132	0.013	Error		



Peak table for sample 4 : bc1028

Peak	Size [bp]	Conc. [pg/ μ l]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	19.1	Lower Marker	0.9	7.6
2	48	193.66	6,158.1	16.3		1.9	18.3
3	59	84.35	2,181.9	8.5		1.1	8.3
4	80	1,281.10	24,122.0	170.5		3.5	132.9
5	966	10.14	15.9	5.4		0.7	3.7
6	1,266	32.45	38.8	10.3		1.4	12.5
7	1,657	86.91	79.5	17.9		2.4	36.2
8	1,831	55.02	45.5	21.6		1.1	23.6
9	1,936	46.06	36.0	24.0		0.8	20.2
10	2,463	829.91	510.5	400.8		2.6	372.5
11	3,547	354.46	151.4	168.1		2.0	162.1
12	5,543	39.22	10.7	21.1		0.8	18.1
13	6,720	24.14	5.4	16.0		0.8	11.2
14	8,308	11.06	2.0	9.2		0.6	5.4
15	9,223	12.89	2.1	8.9		0.7	6.4
16	10,380	75.00	10.9	34.5	Upper Marker	2.2	38.5

bc1028-0

GGAGAA

AAGGAGACTTCGGCTACCCAGAGAAGTTCAGTGCCAGCTCTACTGAGAAGAATGCTGTG
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bc1028-1

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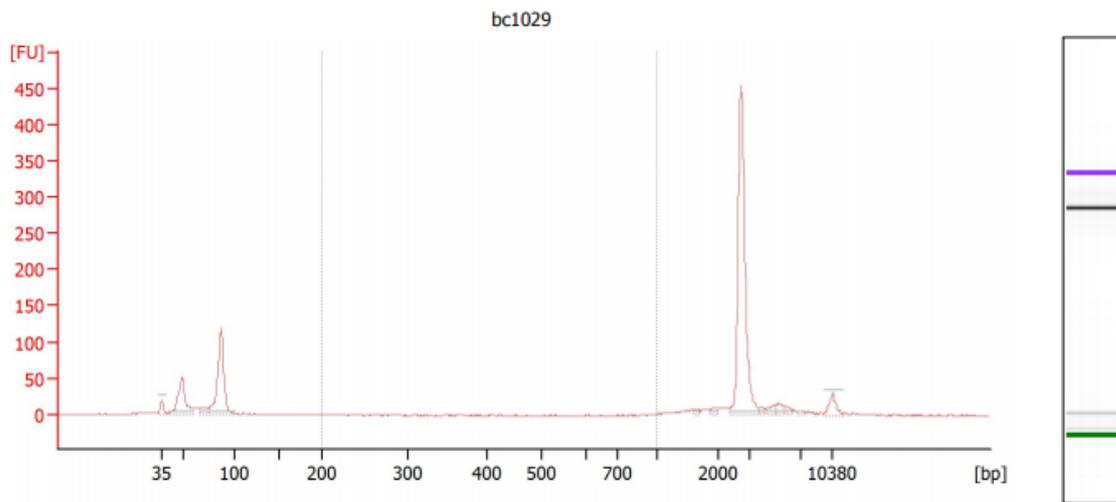
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SC515 / bc1029

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1029-0	2670	6097	0.610	Correct (homozygous)	-	36
1029-1	2671	329	0.033	Error		
1029-2	2671	310	0.031	Error		
1029-3	2671	278	0.028	Error		

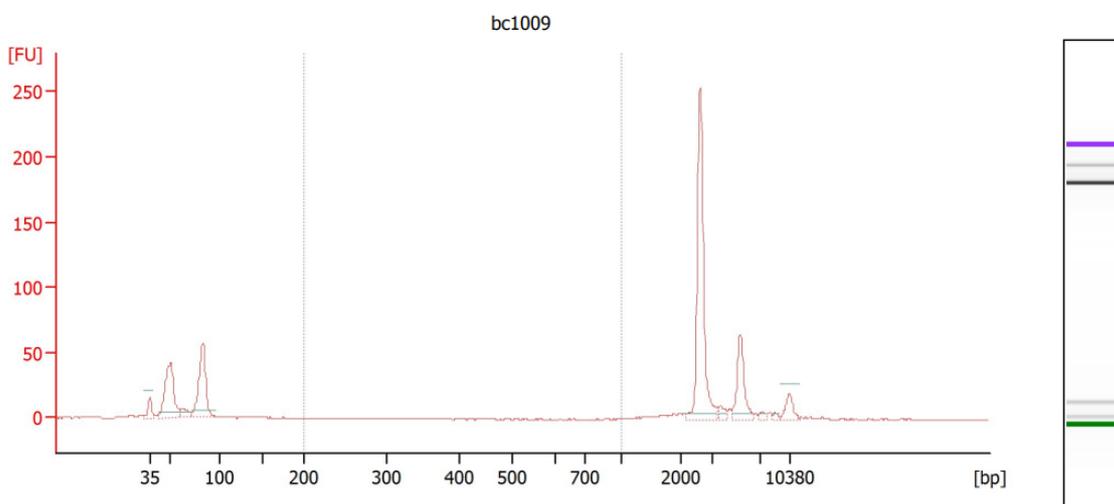


Peak table for sample 2 : **bc1029**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	19.1	Lower Marker	0.9	9.4
2	49	751.65	23,154.6	51.1		2.5	51.5
3	73	132.98	2,776.8	9.7		1.0	9.7
4	87	1,332.06	23,329.7	119.7		2.9	99.9
5	1,625	18.46	17.2	7.9		0.7	5.4
6	1,946	26.96	21.0	10.5		0.8	8.4
7	2,729	1,323.14	734.7	455.7		3.1	424.0
8	3,888	22.23	8.7	11.9		0.6	7.2
9	4,988	39.41	12.0	14.4		1.0	12.9
10	5,263	38.73	11.1	17.2		0.8	12.7
11	5,848	24.90	6.5	12.1		0.7	8.2
12	6,810	14.99	3.3	6.2		0.9	4.9
13	10,380	75.00	10.9	30.6	Upper Marker	2.0	27.3

SC534 / bc1009

Sequence ID	Bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
1009-0	2670	5460	0.546	Correct	-	35
1009-1	2671	450	0.045	Error		
1009-2	2671	432	0.043	Error		
1009-3	2671	350	0.035	Error		
1009-4	5491	267	0.027	Correct	19/C	82
1009-5	2671	168	0.017	Error		
1009-6	2671	149	0.015	Error		



Peak table for sample 5 : bc1009

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	16.5	Lower Marker	1.3	8.2
2	50	965.38	29,120.4	42.0		2.5	50.2
3	63	127.98	3,091.8	6.8		1.2	6.8
4	83	945.97	17,220.5	57.7		2.7	52.9
5	2,623	899.21	519.3	254.9		3.4	213.8
6	3,692	35.30	14.5	10.6		0.9	8.5
7	5,337	256.86	72.9	64.8		2.4	62.5
8	7,281	18.82	3.9	6.2		0.9	4.7
9	8,653	14.05	2.5	5.6		0.9	3.6
10	10,380	75.00	10.9	20.5	Upper Marker	2.1	20.3

bc1009-0 (complementary sequence)

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GCCCACGGTGTACCTCGGCCCCGGACACCAGGCCGGCCCCGGGCTCCACCGACCCCAA	repeat 35

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GCCCACGGTGTACCTCGGCCCCGGACACCAGGCCGGCCCCGGGCTCCACCGGCCCCGG
GGTCCACCGCCCCCCAGCCAGGGTGTACCTCGGCCCCGGACACCAGGCCGGCCCCGG
GGTCCACCGCCCCCCAGCCATGGTGTACCTCGGCCCCGGACAACAGGCCGCCCTTG
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GTTTCTACTCTGGTGACAACGGC

bc1009-4 (complementary sequence)

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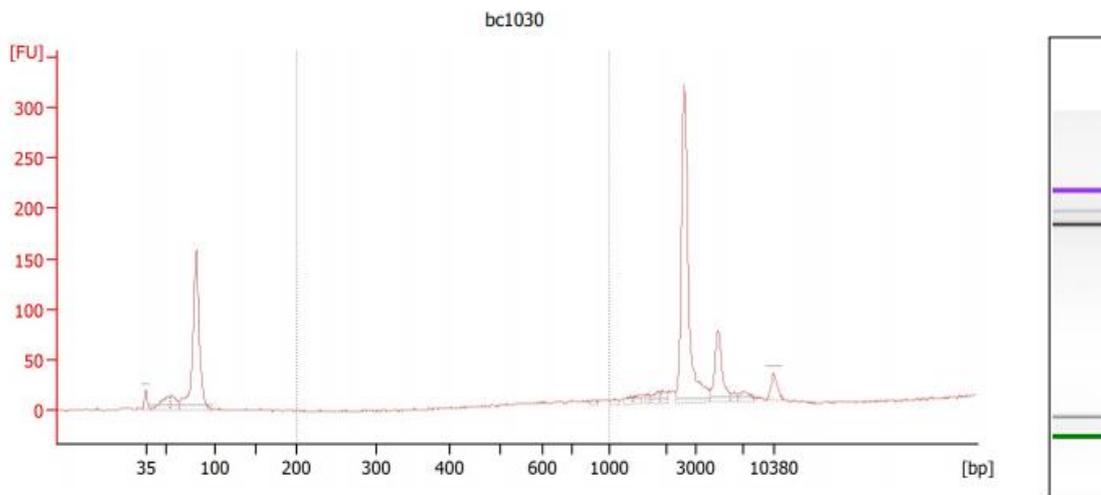
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GCCCACGGTGTACCTCGGCCCGGACACCAGGCCGGCCCCGGGCTCCACCGCCCCCA
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SC560 / bc1030

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1030-0	2670	3335	0.334	Correct	-	36
1030-1	2671	399	0.040	Error		
1030-2	2671	280	0.028	Error		
1030-3	4950	231	0.023	Correct	-	73



Peak table for sample 3 : bc1030

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	20.2	Lower Marker	1.0	8.9
2	53	287.09	8,261.3	13.8		2.2	17.3
3	57	198.41	5,304.7	14.8		1.0	12.0
4	81	2,232.00	41,634.2	158.7		3.8	142.9
5	880	17.58	30.3	5.1		0.8	3.7
6	1,086	21.30	29.7	5.3		1.0	4.8
7	1,372	26.11	28.8	8.0		0.9	6.2
8	1,515	31.51	31.5	9.3		0.9	7.7
9	1,666	25.06	22.8	10.1		0.7	6.3
10	1,854	35.86	29.3	12.4		0.9	9.4
11	1,922	30.99	24.4	12.2		0.7	8.2
12	2,623	1,097.51	633.9	315.3		3.9	300.3
13	4,881	255.46	79.3	70.9		2.3	71.4
14	6,224	24.05	5.9	9.6		0.8	6.8
15	6,878	51.04	11.2	9.9		1.9	14.4
16	10,380	75.00	10.9	28.1	Upper Marker	1.7	23.4

bc1030-0

GGAGAA

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GCCCACGGTGTACCTCGGCCCCGGACACCAGCCGGCCCCGGGCTCCACCGCCCCCAA	repeat 3
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bc1030-3

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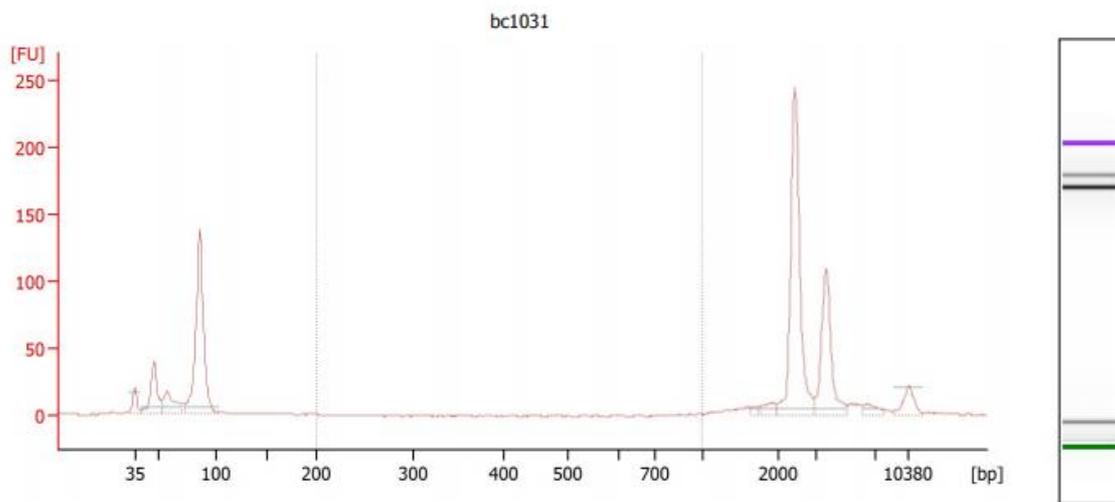
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SC564 / bc1031

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1031-0	2550	4901	0.490	Correct	-	35
1031-1	3870	750	0.075	Correct	-	55
1031-2	1726	141	0.014	Error		
1031-3	2551	128	0.013	Error		



Peak table for sample 6 : **bc1031**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	19.8	Lower Marker	1.0	8.5
2	47	437.64	14,170.6	40.2		1.9	26.6
3	57	304.27	8,093.7	17.3		1.9	19.3
4	85	1,563.91	27,803.4	138.9		3.2	104.9
5	1,719	18.66	16.5	6.8		0.8	4.8
6	1,932	43.45	34.1	9.7		1.5	11.6
7	2,439	813.21	505.1	246.5		3.5	223.6
8	3,639	370.39	154.2	110.6		2.9	103.9
9	6,374	40.89	9.7	8.9		1.9	11.6
10	10,380	75.00	10.9	22.4	Upper Marker	2.6	23.5

bc1031-0

GGAGAA

AAGGAGACTTCGGCTACCCAGAGAAGTTTCAGTGCCCAGCTCTACTGAGAAGAATGCTGTG
AGTATGACCAGCAGCGTACTCTCCAGCCACAGCCCGGTTTCAGGCTCCTCCACCACTCAG
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bc1031-1

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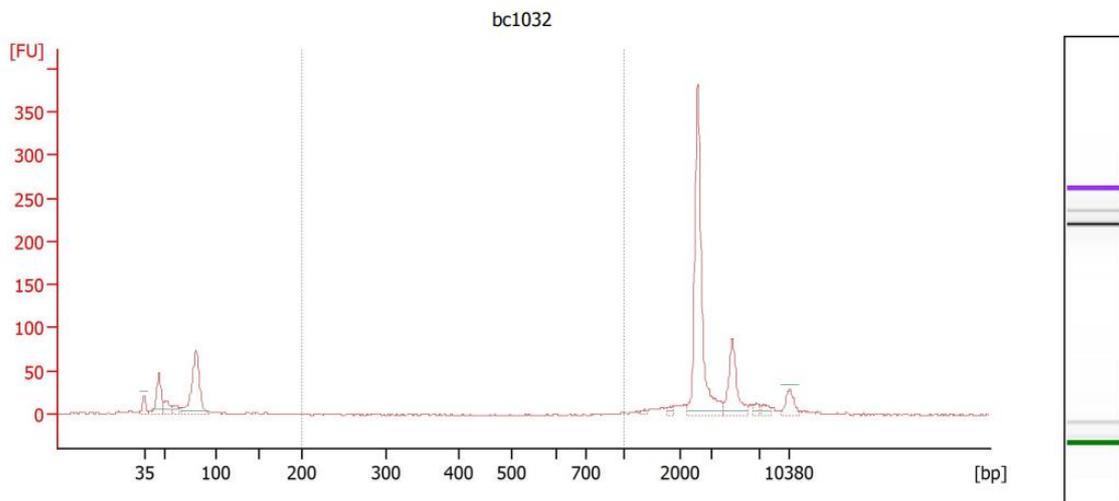
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SC566 / bc1032

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
1032-0	2670	4533	0.453	Correct	-	35
1032-1	2671	502	0.050	Error		
1032-2	4951	461	0.046	Correct	40/C	73
1032-3	2671	307	0.031	Error		



Peak table for sample 5 : bc1032

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	21.3	Lower Marker	0.9	9.8
2	45	384.32	12,821.6	48.7		1.3	30.9
3	51	124.35	3,658.8	15.6		0.8	10.5
4	60	76.41	1,926.0	9.4		0.7	6.6
5	80	882.47	16,639.0	75.2		3.1	79.5
6	1,382	13.50	14.8	6.4		1.0	4.6
7	1,860	19.33	15.7	11.1		0.7	7.2
8	2,566	910.04	537.4	384.3		4.0	354.2
9	4,795	238.50	75.4	88.4		2.6	94.9
10	6,765	23.28	5.2	14.7		0.7	9.4
11	7,421	30.20	6.2	12.5		1.1	12.3
12	10,380	75.00	10.9	29.5	Upper Marker	2.1	33.3

bc1032-0

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bc1032-2

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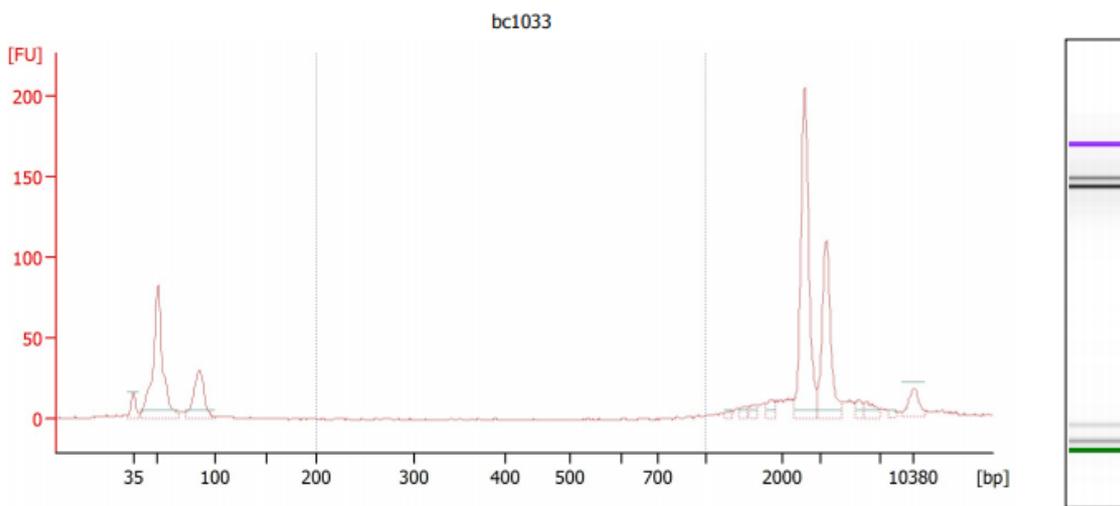
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SC568 / bc1033

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1033-0	2670	3410	0.341	Correct	-	35
1033-1	3570	958	0.096	Correct	-	50
1033-2	2671	235	0.024	Error		
1033-3	3571	232	0.023	Error		



Peak table for sample 7 : bc1033

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	15.8	Lower Marker	1.0	7.3
2	50	1,544.44	46,671.1	82.5		3.4	76.9
3	86	563.92	9,908.9	29.0		2.7	30.1
4	1,340	15.21	17.2	5.1		0.7	2.9
5	1,506	24.56	24.7	7.4		0.8	4.8
6	1,617	26.32	24.7	8.7		0.7	5.3
7	1,847	37.97	31.2	11.2		0.8	7.9
8	2,596	727.13	424.4	205.1		2.1	159.3
9	3,358	419.46	189.2	110.1		2.2	93.2
10	5,520	33.68	9.2	11.1		0.8	7.6
11	6,009	47.23	11.9	10.0		1.5	10.6
12	8,135	13.26	2.5	5.0		0.8	3.1
13	10,380	75.00	10.9	18.1	Upper Marker	2.1	18.6

bc1033-1

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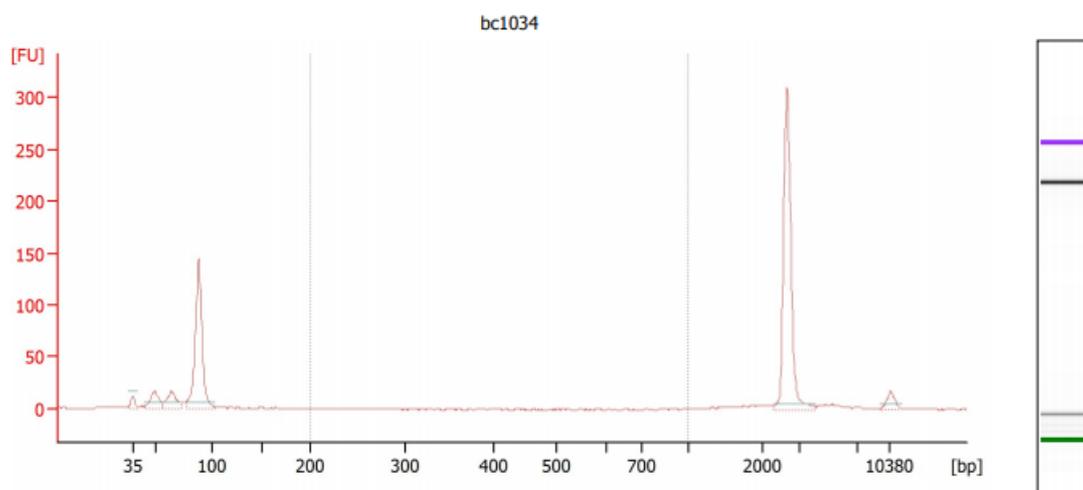
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SC586 / bc1034

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1034-0	2670	2842	0.284	Correct	-	35
1034-1	2670	2314	0.231	Correct	-	35
1034-2	2670	491	0.049	Error		
1034-3	2670	395	0.040	Error		



Peak table for sample 8 : **bc1034**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	11.8	Lower Marker	1.0	4.8
2	49	370.54	11,495.3	16.2		1.7	13.5
3	63	394.19	9,446.2	16.2		1.7	14.9
4	88	2,395.10	41,395.7	144.3		2.8	95.1
5	2,662	1,394.33	793.7	311.6		3.9	226.1
6	10,380	75.00	10.9	17.6	Upper Marker	2.1	13.8

bc1034-0

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bc1034-1

GGAGAA

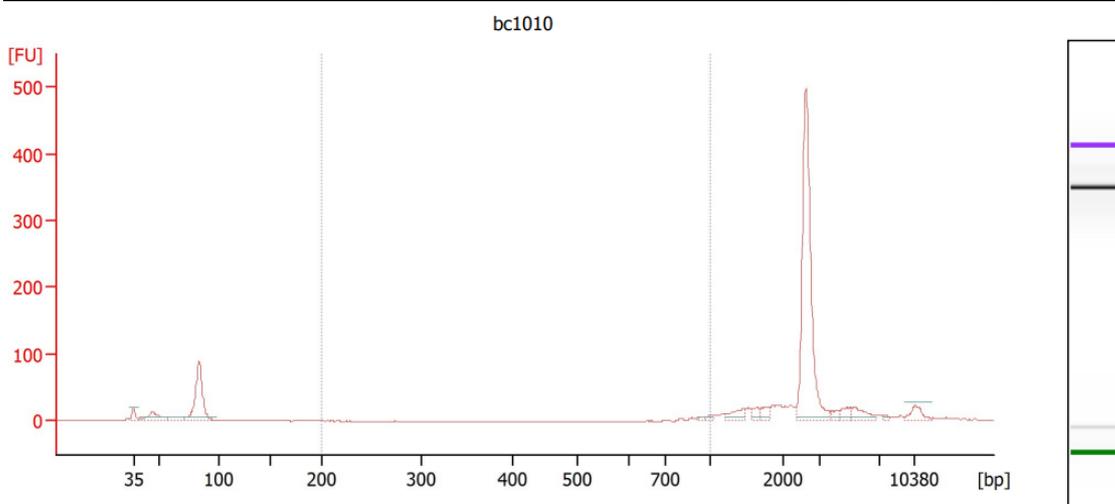
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SC591 / bc1010

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1010-0	2670	4383	0.438	Correct	-	35
1010-1	2670	1975	0.198	Correct	-	35
1010-2	2671	179	0.018	Error		
1010-3	2671	164	0.016	Error		



Peak table for sample 6 : bc1010

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	17.7	Lower Marker	0.9	7.9
2	47	266.97	8,694.5	14.0		2.5	19.1
3	67	127.84	2,889.3	7.0		1.5	9.8
4	83	879.67	15,977.1	91.1		2.9	70.0
5	939	13.04	21.0	6.0		0.7	3.5
6	1,004	17.84	26.9	7.9		0.7	4.9
7	1,450	88.26	92.2	18.7		1.8	26.4
8	1,639	43.60	40.3	19.8		0.7	13.5
9	1,768	52.04	44.6	21.8		0.8	16.5
10	2,608	1,331.67	773.6	499.3		3.0	448.9
11	4,097	28.42	10.5	15.8	Upper Marker	0.6	9.7
12	4,844	58.64	18.3	20.0		1.1	20.2
13	5,291	90.86	26.0	21.2		2.3	31.4
14	7,426	12.94	2.6	8.5		0.6	4.6
15	10,380	75.00	10.9	23.6		2.5	28.7

bc1010-0 (complementary sequence)

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bc1010-1 (complementary sequence)

GGAGAA

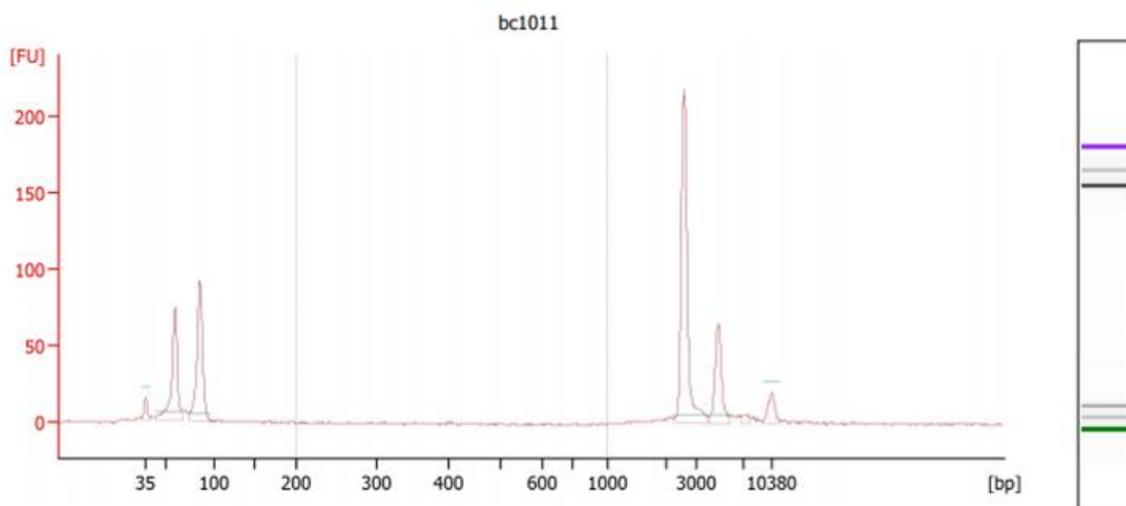
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GGCTCCACCGCCCCCTCAGTCCACAATGTACCTCGGCTCAGGCTCTGCATCAGGCTCA
GCTTCTACTCTGGTGACAACGGC

SC593 / bc1011

Sequence ID	Bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1011-0	2610	883	0.088	Correct	-	34
1011-1	2611	417	0.042	Error		
1011-2	2611	285	0.029	Error		
1011-3	2612	284	0.028	Error		
1011-4	2611	204	0.020	Error		
1011-5	2611	199	0.020	Error		
1011-6	4830	161	0.016	Correct	-	71
1011-7	2611	129	0.013	Error		



Peak table for sample 7 : **bc1011**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	14.5	Lower Marker	0.9	6.0
2	60	1,193.12	30,276.0	74.4		3.1	55.7
3	86	1,387.90	24,584.2	91.9		2.5	68.5
4	2,613	857.08	496.9	218.2		3.8	177.9
5	4,933	263.53	80.9	64.6		2.2	55.9
6	7,070	20.33	4.4	5.6		0.9	4.4
7	10,380	75.00	10.9	20.9	Upper Marker	1.8	17.7

bc1011-0

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bc1011-6

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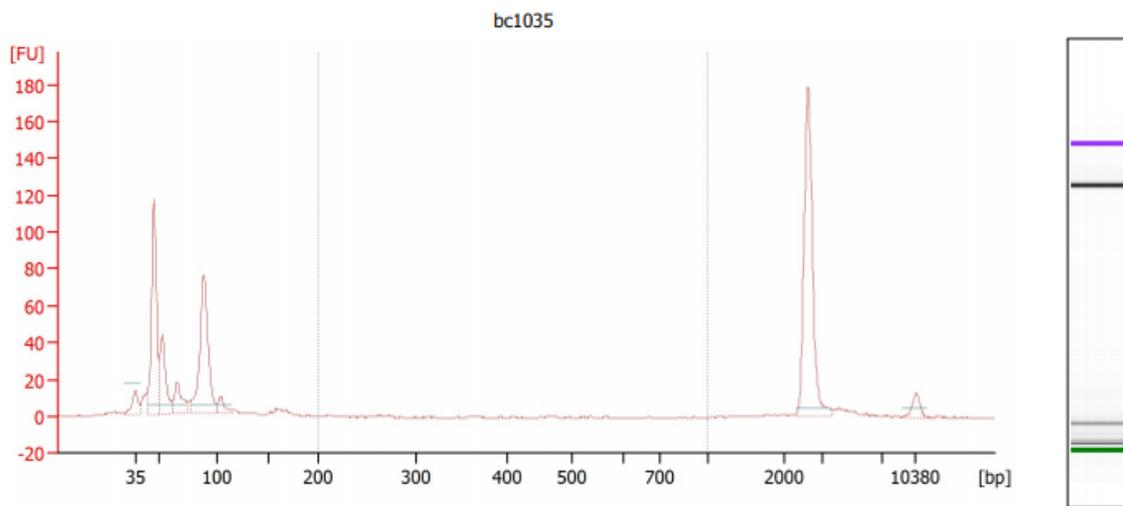
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SC596 / bc1035

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1035-0	2610	4127	0.413	Correct	-	34
1035-1	2730	2272	0.227	Correct	-	36
1035-2	2731	577	0.058	Error		
1035-3	2731	275	0.028	Error		



Peak table for sample 9 : **bc1035**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	12.6	Lower Marker	1.4	7.2
2	47	2,240.91	72,811.0	116.6		1.0	56.7
3	52	908.24	26,390.7	43.2		1.2	23.8
4	65	464.02	10,827.3	17.2		1.3	12.5
5	88	2,151.88	37,113.4	75.8		2.4	60.4
6	103	202.01	2,966.2	9.1		1.3	5.8
7	2,622	1,280.67	740.1	179.3		3.3	146.2
8	10,380	75.00	10.9	12.7	Upper Marker	2.3	9.7

bc1035-0

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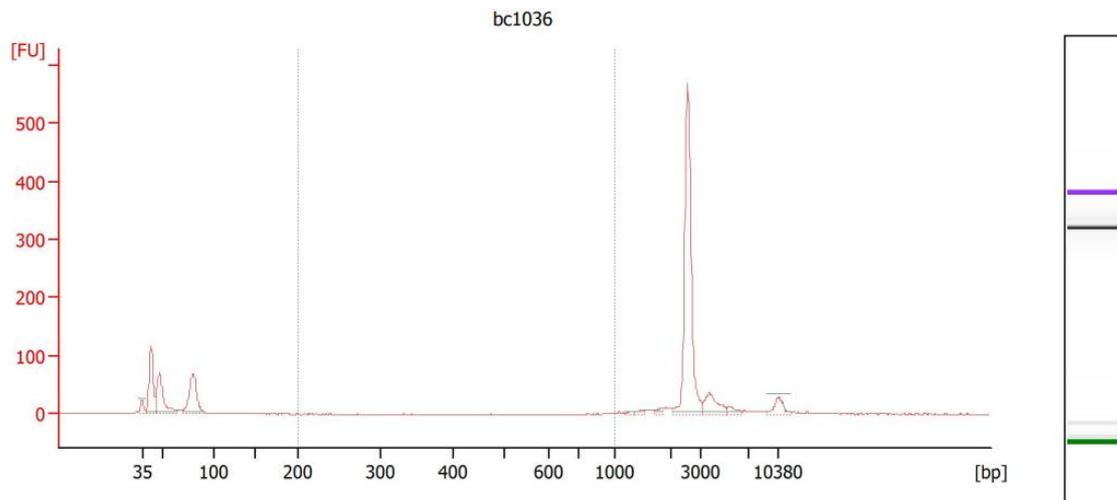
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SC611 / bc1036

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1036-0	2670	2877	0.288	Correct	-	35
1036-1	2610	1443	0.144	Correct	-	34
1036-2	2613	390	0.039	Error		
1036-3	2611	285	0.029	Error		



Peak table for sample 6 : **bc1036**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	23.9	Lower Marker	0.9	11.2
2	41	725.97	26,569.0	116.6		1.1	62.6
3	47	703.95	22,464.4	70.8		2.4	64.3
4	80	706.51	13,349.7	69.8		2.6	70.9
5	1,324	13.19	15.1	6.1		1.1	5.0
6	1,488	19.08	19.4	8.4		1.1	7.4
7	1,816	20.35	17.0	10.7		0.9	8.4
8	2,568	1,146.64	676.5	570.7		3.2	495.7
9	3,732	146.33	59.4	37.5		2.7	64.3
10	5,396	37.86	10.6	13.8		1.7	16.8
11	10,380	75.00	10.9	30.1	Upper Marker	2.8	37.0

bc1036-0

GGAGAA

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GGAGAA

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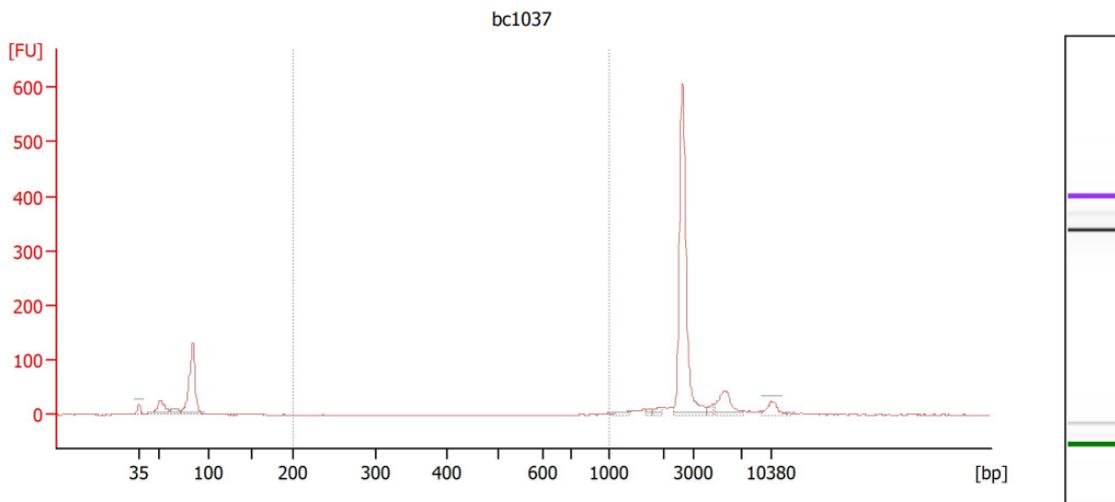
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SC615 / bc1037

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1037-0	2670	4369	0.437	Correct	-	35
1037-1	2610	2760	0.276	Correct	-	34
1037-2	2612	245	0.025	Error		
1037-3	2671	202	0.020	Error		



Peak table for sample 7 : bc1037

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	20.3	Lower Marker	0.9	8.5
2	52	333.57	9,802.0	26.5		2.5	29.6
3	64	117.78	2,773.7	10.3		1.1	10.8
4	84	1,103.74	19,972.6	130.7		2.6	105.6
5	1,347	31.06	34.9	7.7		1.9	11.1
6	1,702	22.11	19.7	12.0		0.7	8.4
7	1,915	36.17	28.6	15.6		1.0	14.3
8	2,615	1,350.01	782.2	609.0		3.7	551.5
9	4,650	35.69	11.6	20.9		0.8	14.9
10	5,582	188.88	51.3	45.6		3.1	79.1
11	10,380	75.00	10.9	25.9	Upper Marker	2.4	34.9

bc1037-0

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bc1037-1

GGAGAA

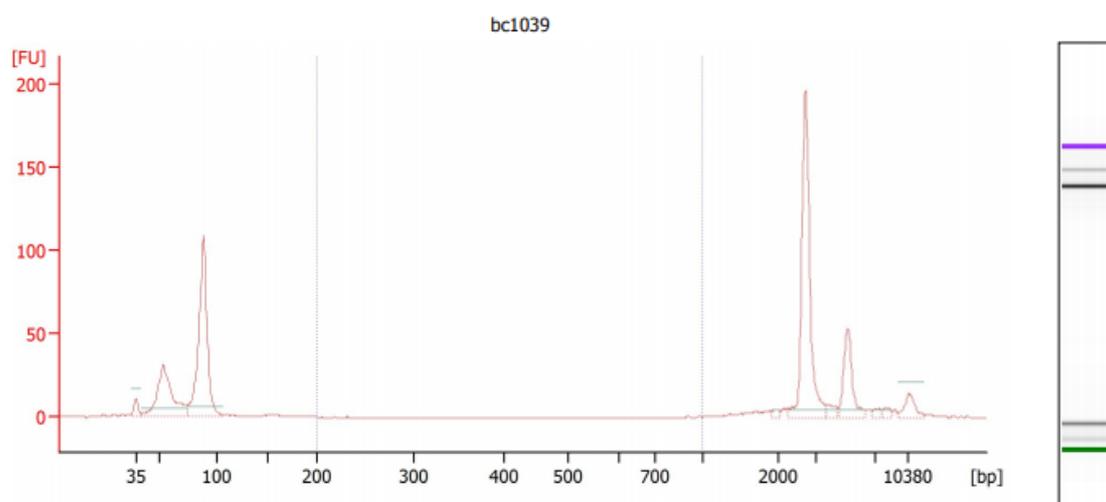
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SC616 / bc1039

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1039-0	2730	4618	0.462	Correct	-	35
1039-1	2731	363	0.036	Error		
1039-2	4950	327	0.033	Correct	-	73
1039-3	2731	223	0.022	Error		



Peak table for sample 10 : bc1039

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	10.9	Lower Marker	0.9	4.4
2	53	1,087.09	31,211.4	30.6		4.0	42.7
3	88	2,063.94	35,501.9	107.9		3.3	86.7
4	1,990	19.90	15.2	5.1		0.8	3.3
5	2,721	882.53	491.4	196.8		3.4	150.8
6	3,848	36.68	14.4	8.1		1.0	6.4
7	5,141	255.51	75.3	54.0		2.4	44.5
8	7,400	22.28	4.6	6.0		0.9	4.0
9	7,897	23.64	4.5	6.0		0.9	4.3
10	10,380	75.00	10.9	14.5	Upper Marker	2.4	14.5

bc1039-0

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bc1039-2

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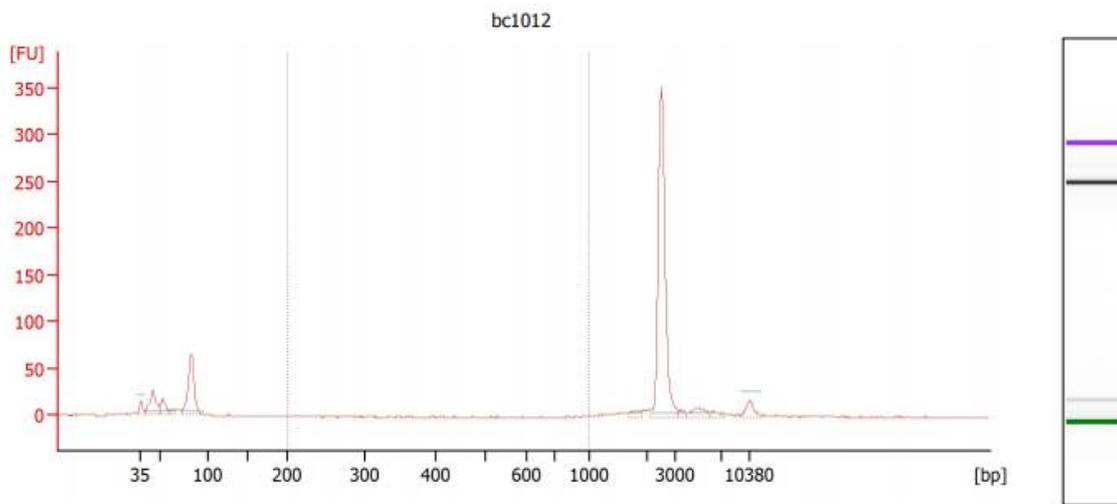
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SC617 / bc1012

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1012-0	2490	3136	0.314	Correct	-	32
1012-1	2610	1035	0.104	Correct	-	34
1012-2	2611	362	0.036	Error	/	/
1012-3	2415	340	0.034	Error	/	/



Peak table for sample 8 : bc1012

Peak	Size [bp]	Conc. [pg/ μ l]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	15.3	Lower Marker	0.9	6.9
2	44	508.01	17,435.1	26.6		1.7	24.1
3	53	250.09	7,192.9	17.6		1.1	12.6
4	64	76.66	1,804.0	6.0		0.7	4.0
5	83	1,015.03	18,503.6	66.2		2.7	54.7
6	1,849	43.63	35.8	7.1		1.5	9.5
7	2,523	1,454.63	873.4	354.7		3.2	329.1
8	3,521	24.96	10.7	7.6		0.9	5.7
9	4,864	80.44	25.1	9.9		2.5	18.6
10	6,170	32.72	8.0	6.6		1.6	7.6
11	10,380	75.00	10.9	18.9	Upper Marker	2.3	19.3

bc1012-0

GGAGAA

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bc1012-1

GGAGAA

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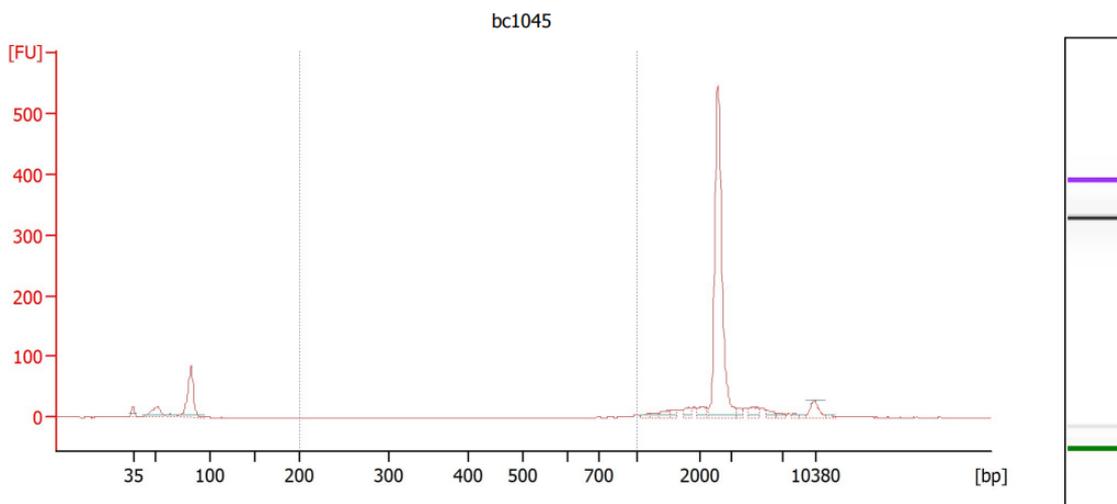
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GCTTCTACTCTGGTGCACAACGGC

SC629 / bc1045

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1045-0	2610	1726	0.173	Correct	-	34
1045-1	2610	1591	0.159	Correct	-	34
1045-2	2611	409	0.041	Error	/	/
1045-3	2611	355	0.036	Error	/	/



Peak table for sample 2 : bc1045

Peak	Size [bp]	Conc. [pg/ μ l]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	17.4	Lower Marker	0.9	7.4
2	52	255.20	7,441.3	16.5		2.3	20.9
3	63	66.68	1,591.4	5.3		1.2	5.6
4	83	704.92	12,849.2	85.6		2.6	62.1
5	1,157	18.72	24.5	7.0		1.0	5.9
6	1,326	21.30	24.3	9.1		0.8	7.0
7	1,489	36.06	36.7	12.5		1.0	12.1
8	1,607	28.22	26.6	13.9		0.7	9.7
9	1,828	42.11	34.9	17.8		0.9	15.1
10	2,112	46.96	33.7	18.4		0.9	17.4
11	2,578	1,317.57	774.5	547.6		2.8	495.8
12	3,564	33.56	14.3	17.0		0.8	12.8
13	4,729	57.66	18.5	18.9		1.2	22.2
14	5,789	29.09	7.6	13.4		1.0	11.3
15	6,601	20.62	4.7	8.7		1.0	8.0
16	8,198	11.04	2.0	7.6		0.7	4.5
17	9,168	9.19	1.5	6.2		0.7	3.8
18	10,380	75.00	10.9	28.1	Upper Marker	2.1	32.2

bc1045-0

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bc1045-1

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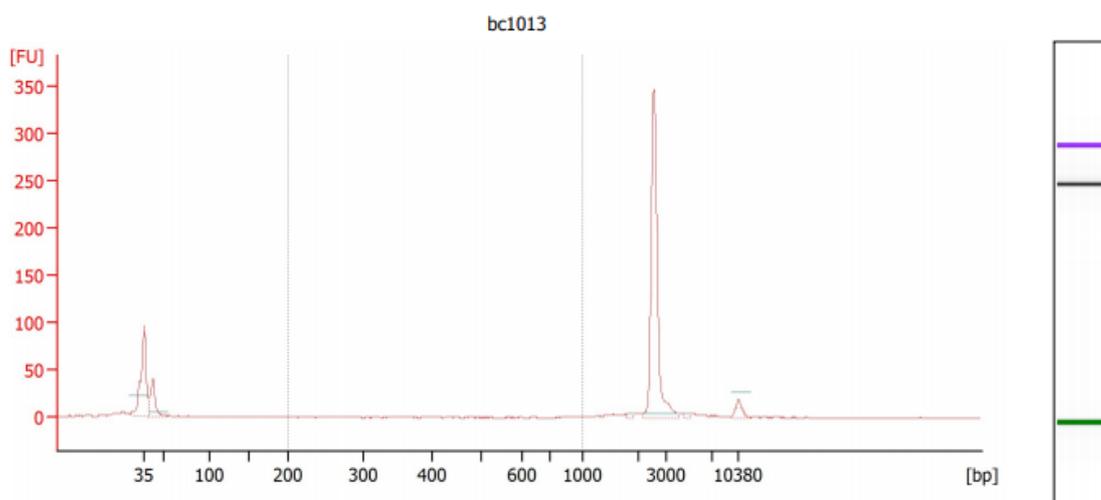
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GCTTCTACTCTGGTGACAACGGC

SC630 / bc1013

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1013-0	2670	2438	0.244	Correct	-	35
1013-1	2610	1217	0.122	Correct	-	34
1013-2	2611	940	0.094	Error		
1013-3	2671	378	0.038	Error		

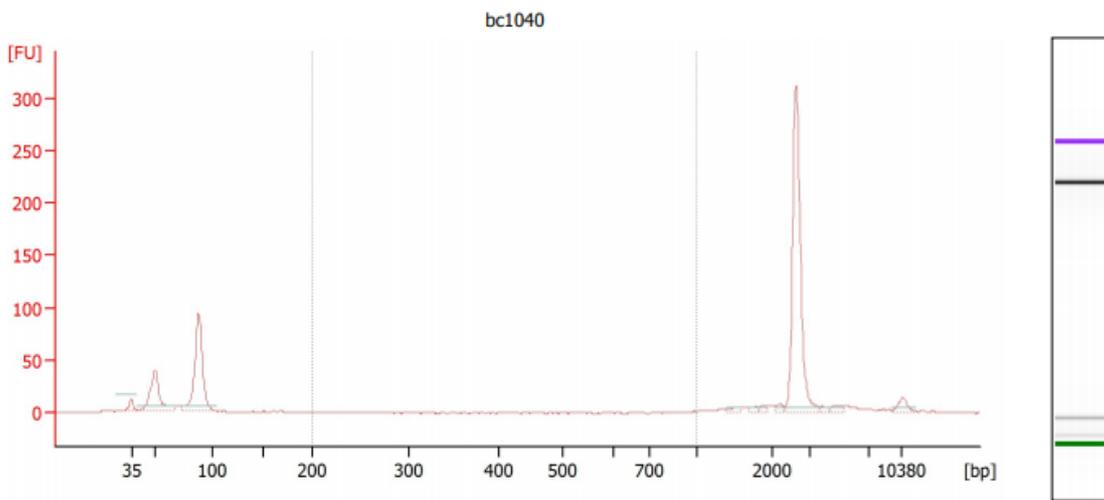


Peak table for sample 9 : **bc1013**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	95.2	Lower Marker	2.3	67.7
2	42	598.45	21,656.3	39.2		2.3	25.2
3	1,861	16.48	13.4	5.1		0.7	3.2
4	2,558	1,457.32	863.1	348.2		4.2	294.1
5	4,762	16.18	5.1	5.5		0.7	3.3
6	10,380	75.00	10.9	19.6	Upper Marker	2.3	17.2

SC634 / bc1040

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1040-0	2670	4661	0.466	Correct (homozygous)	-	35
1040-1	2671	371	0.037	Error		
1040-2	2671	363	0.036	Error		
1040-3	2671	326	0.033	Error		

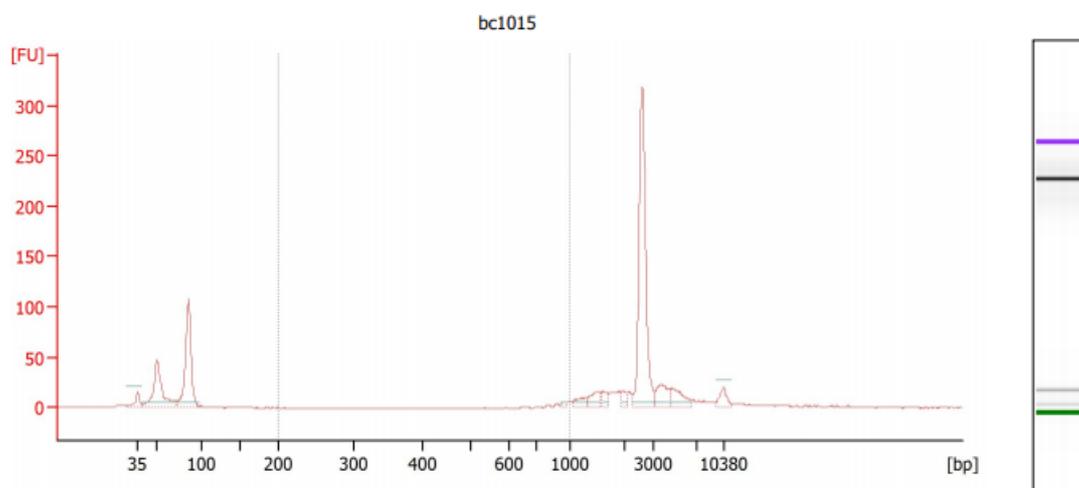


Peak table for sample 11 : **bc1040**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	11.0	Lower Marker	1.9	5.3
2	50	985.26	30,006.4	39.5		3.3	36.9
3	87	1,759.37	30,553.7	93.3		3.3	70.9
4	1,576	38.79	37.3	5.6		1.4	5.8
5	1,796	27.83	23.5	6.2		0.8	4.3
6	1,885	29.55	23.7	7.1		0.8	4.6
7	2,204	33.84	23.3	7.9		0.8	5.5
8	2,632	1,455.03	837.7	313.1		3.1	238.2
9	3,785	27.19	10.9	6.7		0.9	4.5
10	5,083	45.72	13.6	7.3		1.3	7.6
11	10,380	75.00	10.9	14.3	Upper Marker	2.2	13.9

SC638 / bc1015

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1015-0	2610	2222	0.222	Correct	-	34
1015-1	2730	2175	0.218	Correct	-	36
1015-2	2535	368	0.037			
1015-3	2731	266	0.027			



Peak table for sample 10 : bc1015

Peak	Size [bp]	Conc. [pg/ μ l]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	15.2	Lower Marker	1.7	8.7
2	50	1,079.66	32,398.2	47.6		4.2	55.4
3	85	1,493.78	26,504.3	107.0		2.7	82.6
4	948	18.00	28.8	5.6		0.8	3.3
5	1,285	77.34	91.2	10.5		1.8	15.4
6	1,521	99.75	99.4	16.2		1.6	20.7
7	1,607	55.82	52.6	15.7		0.8	11.8
8	1,960	51.06	39.5	16.2		0.8	11.5
9	2,639	1,262.94	725.1	318.7		2.7	292.1
10	3,671	152.41	62.9	22.6		1.8	35.8
11	4,792	141.28	44.7	19.2		2.5	33.3
12	10,380	75.00	10.9	20.0	Upper Marker	1.9	19.7

bc1015-1

GGAGAA

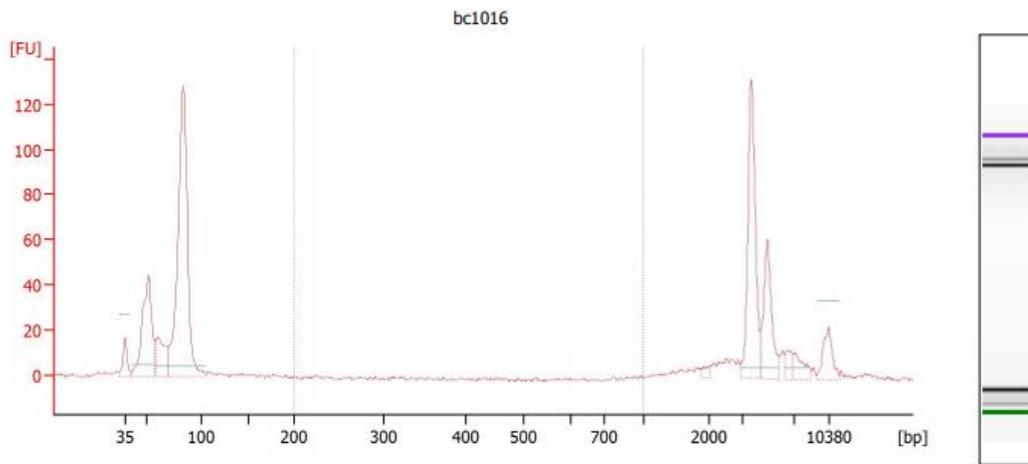
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SC639 / bc1016

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1016-0	3810	2208	0.221	Correct	-	54
1016-1	3811	528	0.053	Error	/	/
1016-2	4950	521	0.052	Correct	-	73
1016-3	4951	224	0.022	Error	/	/



Peak table for sample 6 : **bc1016**

Peak	Size [bp]	Conc. [pg/ μ l]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	17.3	Lower Marker	1.1	8.3
2	52	769.68	22,600.8	44.6		2.4	47.1
3	60	291.76	7,360.4	17.5		1.2	18.2
4	83	2,083.48	37,992.6	129.2		3.7	136.6
5	1,985	16.41	12.5	5.5		0.9	4.4
6	3,694	392.82	161.1	132.9		1.9	109.6
7	4,880	195.85	60.8	61.9		1.7	54.9
8	6,502	30.02	7.0	12.6		0.7	8.5
9	7,021	48.65	10.5	11.7		1.8	13.8
10	10,380	75.00	10.9	23.1	Upper Marker	2.2	23.4

bc1016-0

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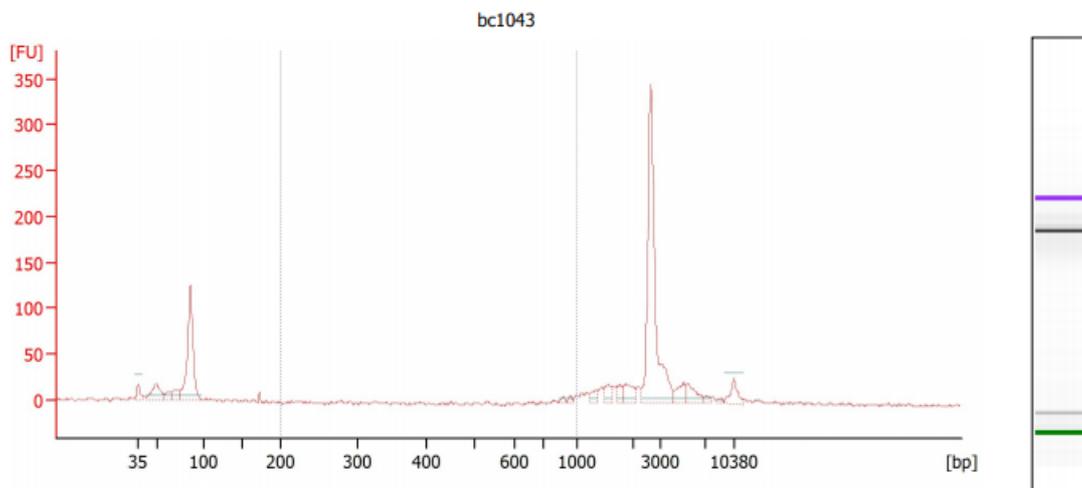
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repeat 43

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repeat 49
repeat 50
repeat 51
repeat 52
repeat 53
repeat 54

SC650 / bc1043

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1043-0	2730	3369	0.337	Correct	-	36
1043-1	2670	1337	0.134	Correct	-	35
1043-2	2670	420	0.042	Error		
1043-3	2059	236	0.024	Error		

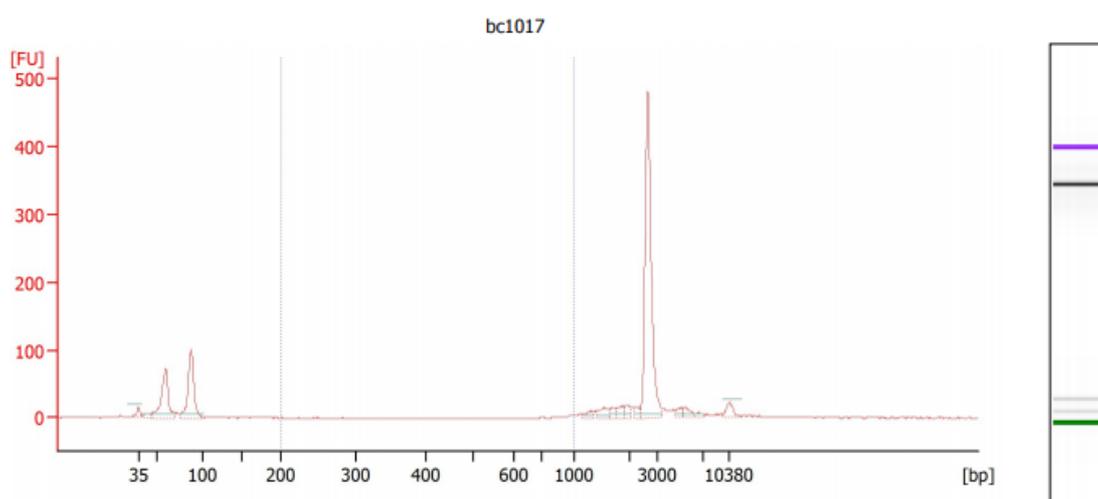


Peak table for sample 9 : **bc1043**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	17.6	Lower Marker	0.9	7.7
2	49	300.41	9,257.4	17.6		2.2	20.9
3	62	84.96	2,067.2	8.4		0.8	6.1
4	69	128.91	2,841.0	10.7		1.0	9.4
5	86	1,241.24	21,904.4	125.5		2.6	94.1
6	875	14.27	24.7	6.0		1.0	3.4
7	938	14.56	23.5	7.4		0.8	3.6
8	1,338	47.36	53.6	14.2		1.1	13.1
9	1,564	59.84	58.0	20.4		1.0	17.3
10	1,775	43.16	36.8	19.8		0.7	12.9
11	1,876	90.30	72.9	20.5		1.5	27.6
12	2,669	1,097.29	623.0	348.5		3.9	349.3
13	5,054	78.08	23.4	23.3		1.4	25.4
14	5,393	90.51	25.4	21.3		2.1	29.5
15	7,115	19.65	4.2	8.7		0.9	6.5
16	8,850	11.46	2.0	5.6		0.8	4.0
17	10,380	75.00	10.9	28.2	Upper Marker	2.4	27.1

SC651 / bc1017

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1017-0	2670	2462	0.246	Correct	-	35
1017-1	2670	2142	0.214	Correct	-	35
1017-2	2671	310	0.031	Error	/	/
1017-3	2671	248	0.025	Error	/	/



Peak table for sample 11 : bc1017

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	15.7	Lower Marker	1.6	9.2
2	44	104.46	3,601.2	6.9		1.0	5.6
3	59	1,213.71	31,040.5	72.0		2.9	69.8
4	87	1,320.75	23,044.0	101.5		2.9	80.5
5	1,311	47.13	54.5	10.2		1.3	10.3
6	1,548	86.16	84.3	15.6		1.5	19.7
7	1,706	39.71	35.3	15.1		0.7	9.4
8	1,872	64.41	52.1	17.6		1.0	15.7
9	1,935	51.14	40.0	17.4		0.8	12.6
10	2,274	40.54	27.0	16.3		0.7	10.2
11	2,660	1,578.27	899.2	482.2		2.7	400.5
12	5,064	44.02	13.2	15.7		0.9	11.4
13	5,454	54.71	15.2	15.1		1.4	14.2
14	6,624	25.45	5.8	7.0		1.2	6.6
15	10,380	75.00	10.9	21.5	Upper Marker	2.2	21.6

bc1017-1

GGAGAA

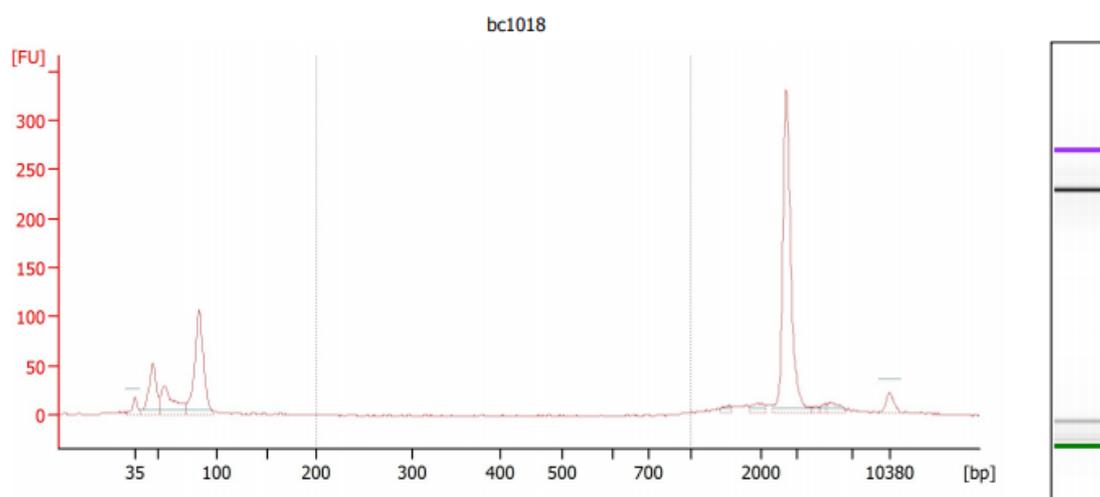
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SC656 / bc1018

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation (repeat / inserted nucleotide)	repeats
1018-0	2731	3271	0.327	Correct	7/C	36
1018-1	2730	1556	0.156	Correct	-	36
1018-2	2732	803	0.080	Error		
1018-3	2732	457	0.046	Error		

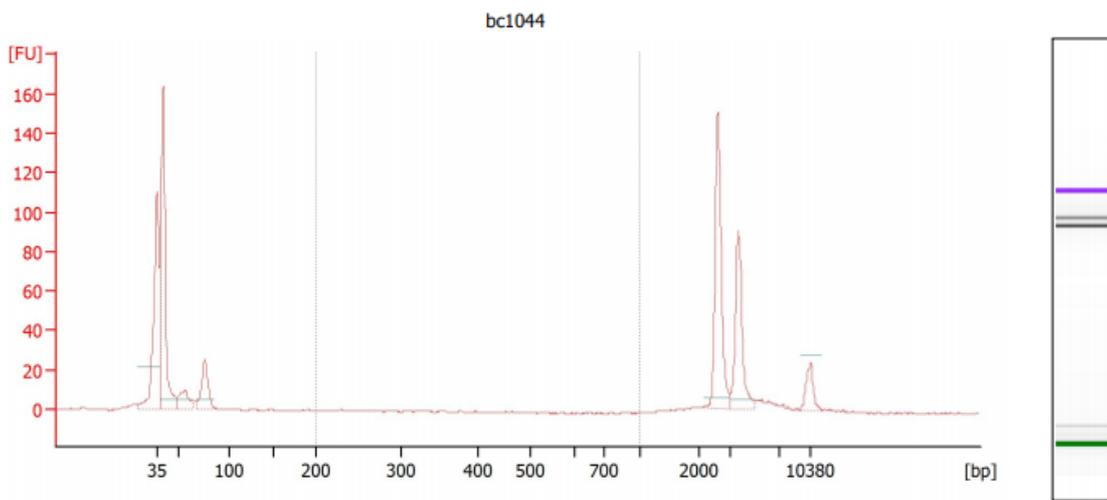


Peak table for sample 7 : bc1018

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	17.9	Lower Marker	1.4	9.8
2	46	922.89	30,273.5	53.2		1.8	42.5
3	55	798.93	21,888.4	29.5		2.3	38.5
4	84	1,952.24	35,029.2	107.3		2.7	100.1
5	1,524	39.90	39.7	8.3		1.1	7.7
6	1,947	64.61	50.3	11.2		1.5	13.4
7	2,683	1,348.00	761.3	330.9		3.6	288.0
8	4,361	20.56	7.1	7.4		0.7	4.5
9	4,964	27.93	8.5	9.9		0.7	6.1
10	5,366	67.72	19.1	11.5		1.8	14.8
11	10,380	75.00	10.9	21.6	Upper Marker	2.2	18.2

SC673 / bc1044

Sequence ID	bp	reads	Allele Frequency	Correct / Error	Mutation	repeats
1044-0	2730	1394	0.139	Correct	-	36
1044-1	3870	1059	0.106	Correct	-	55
1044-2	2731	760	0.076	Error		
1044-3	3871	257	0.026	Error		



Peak table for sample 1 : **bc1044**

Peak	Size [bp]	Conc. [pg/μl]	Molarity [pmol/l]	Peak Height	Observations	Peak Width	Area
1	35	125.00	5,411.3	110.8	Lower Marker	2.3	82.3
2	39	1,751.28	67,521.0	164.3		1.8	94.2
3	56	205.14	5,510.7	9.7		1.7	12.4
4	76	351.61	7,023.1	25.5		2.0	22.2
5	2,606	462.98	269.1	150.6		2.9	126.4
6	3,654	296.23	122.8	90.1		2.5	82.1
7	10,380	75.00	10.9	24.9	Upper Marker	2.4	23.3

bc1044-0

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GCCCACGGTGTACCTCGGCCCGGACACCAGGCCGGCCCCGGGCTCCACCGCCCCCCA
GCCCACGGTGTACCTCGGCCCGGACACCAGGCCGGCCCCGGGCTCCACCGCCCCCCA
GCCCACGGTGTACCTCGGCCCGGACACCAGGCCGGCCCCGGGCTCCACCGCCCCCCA

repeat 44
repeat 45
repeat 46
repeat 47
repeat 48
repeat 49
repeat 50
repeat 51
repeat 52
repeat 53
repeat 54
repeat 55

GCCCACGGTGTACCTCGGCCCGGACACCAGGCCGGCCCCGGGCTCCACCGGCCCGG
GGCTCCACCGCCCCCCAGCCACGGTGTACCTCGGCCCGGACACCAGGCCGGCCCCG
GGCTCCACCGCCCCCCAGCCATGGTGTACCTCGGCCCGGACAACAGGCCCGCCTTG
GGCTCCACCGCCCCCTCAGTCCACAATGTCACCTCGGCCTCAGGCTCTGCATCAGGCTCA
GTTCTACTCTGGTGACAACGGC

Supplementary Table S9. Details of the variants detected by SRS

Age	Sex	Gene	cDNA	Amino acid	Mutation	Disease database	Population database	The in-silico evaluations for missense variants				ACMG interpretation			Version of HaloPlex
								PROVEAN	SIFT	Polyphen2	Mutation Taster	Category	Evidence of pathogenicity	Classification	
54	M	<i>UMOD</i>	NM_003361.3: c.193G>A	p.Asp65Asn	Missense	-	-	Neutral	Deleterious	Probably Damaging	Disease Causing	PS2, PM1, PM2, PM5, PM6, PP2, PP3, PP4	3 Moderate 2 Supporting	Likely Pathogenic	4
29	M	<i>UMOD</i>	NM_003361.3: c.739G>C	p.Arg247Pro	Missense	-	-	Deleterious	Deleterious	Probably Damaging	Disease Causing	PM1, PM2, PM5, PP2, PP3, PP4	2 Moderate 3 Supporting	Likely Pathogenic	4
18	M	<i>UMOD</i>	NM_003361.3: c.336C>G	p.Cys112Trp	Missense	-	-	Deleterious	Deleterious	Probably Damaging	Disease Causing	PM1, PM2, PM5, PP2, PP3, PP4	2 Moderate 3 Supporting	Likely Pathogenic	5
62	M	<i>UMOD</i>	NM_003361.3: c.376T>A	p.Cys126Ser	Missense	-	-	Deleterious	Deleterious	Probably Damaging	Disease Causing	PM1, PM2, PM5, PP2, PP3, PP4	2 Moderate 3 Supporting	Likely Pathogenic	5
50	M	<i>UMOD</i>	NM_003361.3: c.175G>T	p.Asp59Tyr	Missense	-	-	Deleterious	Deleterious	Probably Damaging	Disease Causing	PM1, PM2, PM5, PP2, PP3, PP4	3 Moderate 2 Supporting	Likely Pathogenic	6
44	F	<i>UMOD</i>	NM_003361.3: c.601G>C	p.Gly201Arg	Missense	-	rs74753651 0 (dbSNP)	Deleterious	Deleterious	Possibly Damaging	Polymorphism	PM1, PM2, PM5, PP2, PP3, PP4	2 Moderate 3 Supporting	Likely Pathogenic	8
19	F	<i>REN</i>	NM_000537.3: c.58T>G	p.Cys20Gly	Missense	-	-	Deleterious	Deleterious	Benign	Polymorphism	PS1, PM2, PM5, PP3, PP4	2 Moderate 3 Supporting	Likely Pathogenic	4
16	F	<i>NPHP1</i>	NM_00272.5	whole gene deletion of <i>NPHP1</i>								PVS1, PM2, PM4	1 Very Strong 2 Strong	Pathogenic	8

Supplementary Table S10. Clinicopathological characteristics of patients with SRS-detected variants other than *MUC1*

ID	age	sex	gene	FH	renal biopsy	pathological diagnosis/findings	HTN (age at diagnose)	HUA (age at diagnose)	childhood anemia (age at diagnose)	serum potassium (mEq/L)	eGFR	UP
SC395	54	M	<i>UMOD</i>	-	+	IF/TA	-	+(NA)	-	4.3	41.8 (pRTx)	0.01 (ACR)
SC408	29	M	<i>UMOD</i>	+	+	FGO tubulointerstitial scarring	+(28y)	+(NA)	-	4.1	36.4	not detectable
SC430	18	M	<i>UMOD</i>	+	-	-	-	+(NA)	-	4.7	53.9	not detectable
SC447	62	M	<i>UMOD</i>	+	+	MGA	-	+(NA)	-	4.7	43.9	0.05
SC492	50	M	<i>UMOD</i>	+	+	MGA tubulor atropy	+(NA)	+(NA)	-	4.6	35.6	NA
SC660	44	F	<i>UMOD</i>	+	-	-	+(39y)	+(NA)	-	4.1	37.3	0.02 (ACR)
SC317	19	F	<i>REN</i>	+	+	NA	-	+(9y)	+(9y)	4.6	59.7	0.02
SC675	16	F	<i>NPHP1</i>	-	+	chronic TIN	+(NA)	-	+(14y)	3.3	22.5	0.78

FH, family history of CKD; HTN, hypertension; HUA, hyperuricemia; eGFR, estimated glomerular filtration rate (mL/min/1.73 m²); UP, urinary protein (g/gCr); ACR, albumin creatinine ratio; IF/TA; interstitial fibrosis and tubular atrophy; FGO, focal glomerular obsolescence; MGA, minor glomerular abnormalities; TIN, tubulointerstitial nephritis; pRTx; post renal transplantation; NA, not available

Supplementary Table S11. Summary of identifying alleles of each patient analyzed by LRS

Patient ID	sequence ID	bp	reads	Allele frequency	Mutation (repeat / inserted nucleotide)	repeats
SC270	1001-0	2370	3368	0.337	-	30
	1001-1	2670	2977	0.298	-	35
SC298	1002-0	2610	3097	0.31	-	34
	1002-1	2670	2926	0.293	-	35
SC356	1003-0	2731	2895	0.29	10 /C	36
	1003-1	2670	1005	0.101	-	35
SC360	1019-0	2610	3214	0.321	-	34
	1019-1	2670	3036	0.304	-	35
SC362	1020-0	2730	3867	0.387	-	36
	1020-1	2670	3578	0.358	-	35
SC370	1004-0	2731	3250	0.392	5 /G	36
	1004-1	4290	985	0.119	-	62
SC416	1006-0	2670	3889	0.389	-	35
	1006-1	2611	3095	0.31	6/C	34
SC422	1021-0	3870	3967	0.397	-	55
	1021-1	4950	1335	0.134	-	73
SC431	1022-0	2670	3235	0.324	-	35
	1022-1	2610	2812	0.281	-	34
SC449	1023-0	2670	3789	0.379	-	35
	1023-3	4770	548	0.055	-	70
SC465	1024-0	2550	3263	0.326	-	33
	1024-1	2670	2648	0.265	-	35
SC469	1026-0	2670	3339	0.334	-	35
	1026-1	2730	3253	0.325	-	36
SC489	1007-0	2670	3146	0.315	-	35
	1007-1	2731	1785	0.179	7/C	36
SC507	1008-0	2670	3390	0.339	-	35
	1008-1	2670	2567	0.257	-	35
SC511	1027-0	2730	1889	0.189	-	36
	1027-3	4830	163	0.016	-	71
SC512	1028-0	2610	3816	0.382	-	34
	1028-1	3871	874	0.087	14/C	55
SC515	1029-0	2670	6097	0.61	-	35
SC534	1009-0	2670	5460	0.546	-	35
	1009-4	5491	267	0.027	19/C	82
SC560	1030-0	2670	3335	0.334	-	35
	1030-3	4950	231	0.023	-	73
SC564	1031-0	2550	4901	0.49	-	33
	1031-1	3870	750	0.075	-	55
SC566	1032-0	2670	4533	0.453	-	35
	1032-2	4951	461	0.046	40/C	73
SC568	1033-0	2670	3410	0.341	-	35
	1033-1	3570	958	0.096	-	50
SC586	1034-0	2670	2842	0.284	-	35
	1034-1	2670	2314	0.231	-	35

Supplementary Table S11. Summary of identifying alleles of each patient analyzed by LRS (continued)

Patient ID	sequence ID	bp	reads	Allele frequency	Mutation (repeat / inserted nucleotide)	repeats
SC591	1010-0	2670	4383	0.438	-	35
	1010-1	2670	1975	0.198	-	35
SC593	1011-0	2610	883	0.088	-	34
	1011-6	4830	161	0.016	-	71
SC596	1035-0	2610	4127	0.413	-	36
	1035-1	2730	2272	0.227	-	34
SC611	1036-0	2670	2877	0.288	-	35
	1036-1	2610	1443	0.144	-	34
SC615	1037-0	2670	4369	0.437	-	35
	1037-1	2610	2760	0.276	-	34
SC616	1039-0	2730	4618	0.462	-	36
	1039-2	4950	327	0.033	-	73
SC617	1012-0	2490	3136	0.314	-	32
	1012-1	2610	1035	0.104	-	34
SC629	1045-0	2610	1726	0.173	-	34
	1045-1	2610	1591	0.159	-	34
SC630	1013-0	2670	2438	0.244	-	35
	1013-1	2610	1217	0.122	-	34
SC634	1040-0	2670	4661	0.466	-	35
SC638	1015-0	2610	2222	0.222	-	34
	1015-1	2730	2175	0.218	-	36
SC639	1016-0	3810	2208	0.221	-	54
	1016-2	4950	521	0.052	-	73
SC650	1043-0	2730	3369	0.337	-	36
	1043-1	2670	1337	0.134	-	35
SC651	1017-0	2670	2462	0.246	-	35
	1017-1	2670	2142	0.214	-	35
SC656	1018-0	2731	3271	0.327	7/C	36
	1018-1	2730	1556	0.156	-	36
SC673	1044-0	2730	1394	0.139	-	36
	1044-1	3870	1059	0.106	-	55

Supplementary Figure S1. Distribution of repeat number of VNTR of each allele

